## IlluminaHumanMethylation450kanno.ilmn12.hg19

February 11, 2020

 $Illumina Human Methylation 450 kanno.ilmn 12.hg 19\\ Annotation\ data\ for\ the\ `Illumina Human Methylation 450 k'\ micro\ array.$ 

## **Description**

The annotation data is based on Illumina's v1.2 annotation, specifically the file 'HumanMethylation450\_15017482\_v.1.2.csv' derived from 'HumanMethylation450\_15017482\_v.1.2.bpm'. Additional SNP annotation is generated by the authors.

## **Format**

An object of class IlluminaMethylationAnnotation for IlluminaHumanMethylation450kanno.ilmn12.hg19. All others are of class DataFrame

#### **Details**

The following changes/ modifications / addition has been made to the source material.

For the annotation related to UCSC islands, a value of "" has been changed to "OpenSea".

The creation of this object based on the Illumina annotation and additional SNP information (see below) is contained in the createAnno.R script in the scripts directory.

Contents of the columns are the responsibility of Illumina; see their documentation for details.

#### **Source**

See description.

#### **Examples**

data(IlluminaHumanMethylation450kanno.ilmn12.hg19)
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data(Islands.UCSC)

SNPs.CommonSingle SNP annotation from various versions of dbSNP as represented on UCSC Common SNP table.

## **Description**

SNP annotation from various versions of dbSNP as represented on UCSC Common SNP table. Overlap is based on genomic mappings from the annotation package.

## **Format**

An object of class DataFrame. Rownames are CpG identifiers. There are 6 columns Probe\_rs, Probe\_maf, CpG\_rs, CpG\_maf, SBE\_rs, and SBE\_maf. 'Probe' indicates a SNP in the probe, 'CpG' a SNP in the CpG site and 'SBE' in the single base extension site. The \_rs gives the SNP RS identifier and the \_maf gives the minor allele frequency.

#### **Details**

In addition to the SNP information provided by Illumina, we have added independent information on the overlap of the 450k with various versions of dbSNP. The overlap is based on the mappings of the array to the hg19 genome provided by Illumina. As dbSNP we have used the 'Common' table from UCSC (ie. 'snp137Common'). This track contains variants from dbSNP which have a minor allele frequency (MAF) of greater than 1 percent (specifically, this requires dbSNP to actually contain MAF information). Furthermore, we only kept variants marked as 'single' (ie. standard single nucleotide changes, but not insertions or deletions). Scripts for retrieving the UCSC dbSNP table and doing the overlap are contained in the scripts directory. The variants are described in 6 different columns. Probe\_rs tells us the RS number (SNP ID number) for a SNP overlapping the probe, and Probe\_maf is the minor allelle frequency for the SNP (in case multiple SNPs overlap, only one is recorded). Similarly, CpG\_rs describe SNPs overlapping the CpG site and SBE\_rs the single base extension of the measured methylation loci.

## **Source**

UCSC Common SNP Table.

#### **Examples**

```
data(SNPs.147CommonSingle)
data(SNPs.146CommonSingle)
data(SNPs.144CommonSingle)
data(SNPs.142CommonSingle)
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data(SNPs.132CommonSingle)
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