# IlluminaHumanMethylationEPICv2anno.20a1.hg38

March 5, 2025

 $Illumina Human Methylation EPIC v2 anno. 20 a 1.hg 38 \\ Annotation\ data\ for\ the\ `Illumina Human Methylation EPIC'\ micro\ array.$ 

## **Description**

This package is based on the file https://support.illumina.com/content/dam/illumina-support/documents/downloads/productfiles/methylationepic/MethylationEPIC%20v2%20Files.zip from https://support.illumina.com/array/array\_kits/infinium-methylationepic-beadchip-kit/downloads.html.

Additional SNP annotation is generated by the authors (described in SNPs. CommonSingle).

A script for creating the data object in this package is contained in scripts/createAnno.R.

## **Format**

An object of class IlluminaMethylationAnnotation for IlluminaHumanMethylationEPIv2Canno.20a1.hg38. 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 SNPs.CommonSingle) 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**

```
IlluminaHumanMethylationEPICv2anno.20a1.hg38
Locations
Manifest
data(SNPs.Illumina) # data object too large, need to be loaded manually by data()
Islands.UCSC
data(Other) # data object too large, need to be loaded manually by data()
```

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 EPIC (v2.0) array with various versions of dbSNP. The overlap is based on the mappings of the array to the hg38 genome provided by Illumina. As dbSNP we have used the 'Common' table from UCSC (ie. 'snp151Common'). This track contains variants from dbSNP which have a minor allele frequency (MAF) of greater than 1 percent (specifically, this requires db-SNP 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.

aggregate\_to\_probes 3

### **Examples**

```
SNPs.141CommonSingle
SNPs.142CommonSingle
SNPs.144CommonSingle
SNPs.146CommonSingle
SNPs.147CommonSingle
SNPs.150CommonSingle
SNPs.151CommonSingle
```

aggregate\_to\_probes

Aggregate to the probe-level

### **Description**

Aggregate to the probe-level

# Usage

```
aggregate_to_probes(x)
```

# **Arguments**

Х

A data frame or a matrix.

# **Details**

If x is a data frame, it should be one of the following objects: Islands.UCSC, Locations, Other, SNPs.Illumina, SNPs.141CommonSingle, SNPs.142CommonSingle, SNPs.144CommonSingle, SNPs.146CommonSingle, SNPs.151CommonSingle, SNPs.151CommonSingle.

If x is a matrix, it should come from the analysis with the \*\*minfi\*\* package. If multiple rows correspond to the same probe, the average value of rows is simply used.

# **Examples**

```
\label{eq:total_problem} \mbox{\ensuremath{\mbox{\sc H}}} \mbox{\ensuremath{\mbox{\sc There}}} \mbox{\ensuremath{\mbox{\sc is}}} \mbox{\ensuremath{\mbox{\sc NULL}}} \mbox{\
```

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```