## Package 'coMET'

## October 24, 2014

#### Type Package

**Title** coMET: visualisation of regional epigenome-wide association scan (EWAS) results and DNA comethylation patterns.

**Version** 0.99.3

Date 2014-10-17

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## Description

Creates plots of p-values of CpG DNA methylation. Main features of the package include options to display a linkage disequilibrium (LD) plot. Images are created as either PDF/EPS files.

**Depends** R (>= 3.1.0), grid, biomaRt, Gviz, rtracklayer, GenomicRanges

Suggests knitr, RUnit, BiocGenerics, BiocStyle

Imports colortools, hash, grDevices, gridExtra, ggbio, ggplot2, trackViewer

License GPL (>= 2)

URL http://epigen.kcl.ac.uk/comet

#### biocViews

Software, DifferentialMethylation, Visualization, Sequencing, Genetics, FunctionalGenomics

VignetteBuilder knitr

NeedsCompilation no

Repository Bioconductor

2 coMET-package

## **R** topics documented:

coME	T-package coMET: visualisation of regional epigenome-wide association scar (EWAS) results and DNA co-methylation patterns	n
Index		3'
		-
	transcriptENSEMBL	
	•	
	snpLocationsUCSC	
		3
	regulationBiomart	3
	knownGenesUCSC	3
	ISCATrack	
	HistoneOne	
	Histone All	
	genesNameENSEMBL	
	genesENSEMBL	
	GeneReviewsTrack	2
	gcContent	
	GADTrack	
	DNAseUCSC	2
	cpgIslandsUCSC	1
	COSMICTrack	1
	CoreillCNVTrack	1
	comet.web	1
	comet	9
	ClinVarMainTrack	
	ClinVarCnvTrack	
	chrUCSC2ENSEMBL	,
	chromatinHMMOne	
	chromatinHMMAll	
	coMET-package	-

## Description

coMET is an R package for visualising EWAS results in a genomic region. Along with phenotype-association plots, coMET also generates plots of co-methylation patterns and provides a series of annotation tracks.

coMET-package 3

#### **Details**

Package: coMET
Type: Package
Version: 0.99.3
Date: 2014-10-16
License: GPL (>=2)

coMET is an R package that can generate regional plots of EWAS results, DNA co-methylation patterns, and genomic information. A coMET figure includes 3 panels with a plot of P-values from EWAS, customized annotation tracks, and a triangle heatmap plot which demonstrates the correlation structure of DNA methylation at the CpG sites in the genomic region. Plots are created as PDF or EPS files.

#### Author(s)

Tiphaine C. Martin, Idil Yet, Pei-Chien Tsai, Jordana T. Bell Maintainer: Tiphaine Martin <tiphaine.martin@kcl.ac.uk> Website: http://www.epigen.kcl.ac.uk/comet

#### References

Martin, T.C, Erte, I, Tsai, P-C, Bell, J.T., coMET: an R plotting package to visualize regional plots of epigenome-wide association scan results, QG14, 2014.

### **Examples**

```
extdata <- system.file("extdata", package="coMET",mustWork=TRUE)</pre>
configfile <- file.path(extdata, "config_cyp1b1_zoom_4comet.txt")
myinfofile <- file.path(extdata, "cyp1b1_infofile.txt")</pre>
myexpressfile <- file.path(extdata, "cyp1b1_infofile_exprGene_region.txt")</pre>
mycorrelation <- file.path(extdata, "cyp1b1_res37_rawMatrix.txt")</pre>
chrom <- "chr2"
start <- 38290160
end <- 38303219
gen <- "hg19"
if(interactive()){
  genetrack <-genesENSEMBL(gen,chrom,start,end,showId=FALSE)</pre>
  snptrack <- snpBiomart(chrom, start, end,</pre>
                          dataset="hsapiens_snp_som", showId=FALSE)
  strutrack <- structureBiomart(chrom, start, end,</pre>
                                   strand, dataset="hsapiens_structvar_som")
  clinVariant<-ClinVarMainTrack(gen,chrom,start,end)</pre>
  clinCNV<-ClinVarCnvTrack(gen,chrom,start,end)</pre>
  gwastrack <-GWASTrack(gen,chrom,start,end)</pre>
  geneRtrack <-GeneReviewsTrack(gen,chrom,start,end)</pre>
```

4 chromatinHMMAII

```
listgviz <- list(genetrack,snptrack,strutrack,clinVariant,</pre>
                 clinCNV,gwastrack,geneRtrack)
comet(config.file=configfile, MYDATA.FILE=myinfofile, CORMATRIX.FILE=mycorrelation,
      MYDATA.LARGE.FILE=myexpressfile, TRACKS.GVIZ=listgviz,
      VERBOSE=FALSE, PRINT.IMAGE=FALSE, DISP.PVALUEPLOT=FALSE)
} else {
 data(geneENSEMBLtrack)
 data(snpBiomarttrack)
 data(ISCAtrack)
 data(strucBiomarttrack)
 data(ClinVarCnvTrack)
 data(clinVarMaintrack)
 data(GWASTrack)
 data(GeneReviewTrack)
 listgviz <- list(genetrack,snptrack,strutrack,clinVariant,</pre>
                 clinCNV,gwastrack,geneRtrack)
comet(config.file=configfile, MYDATA.FILE=myinfofile, CORMATRIX.FILE=mycorrelation,
      MYDATA.LARGE.FILE=myexpressfile, TRACKS.GVIZ=listgviz,
      VERBOSE=FALSE, PRINT.IMAGE=FALSE, DISP.PVALUEPLOT=FALSE)
}
```

chromatinHMMAll

Creating multiple chromHMM tracks from the UCSC genome browser

## Description

Create multiple chromHMM Broad tracks by connecting to the UCSC genome browser using the GViz bioconductor package

#### Usage

```
chromatinHMMAll(gen, chr, start, end, mySession, track.name = "Broad ChromHMM", pattern = NULL, table.
```

gen	the name of genome
chr	the chromosome of interest
start	the first position in region of interest (the smallest value)
end	the last position in region of interest (the biggest value)
mySession	the object session from the function browserSession of rtracklayer
track.name	the name of the track, for example : Broad ChromHMM
pattern	the pattern of the track to visualise
table.name	the name of the table from the track

chromatinHMMOne 5

#### Value

list of AnnotationTrack objects of GViz

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
 gen <- "hg19"
 chr <- "chr2"
 start <- 38290160
 end <- 38313219
if(interactive()){
 BROWSER.SESSION="UCSC"
 mySession <- browserSession(BROWSER.SESSION)</pre>
 genome(mySession) <- gen</pre>
 track.name="Broad ChromHMM"
 tablestrack<-tableNames(ucscTableQuery(mySession, track=track.name))</pre>
 table.name<-tablestrack[1]</pre>
 PATTERN.REGULATION<-"GM12878"
 chromhmmPattern<-chromatinHMMAll(gen,chr,start,end,mySession,track.name,PATTERN.REGULATION)</pre>
 plotTracks(chromhmmPattern, from = start, to =end)
 chromhmmNoPattern<-chromatinHMMAll(gen,chr,start,end,mySession,track.name)</pre>
 plotTracks(chromhmmNoPattern, from = start, to =end)
} else {
 data(chromhmmPattern)
 plotTracks(chromhmmPattern, from = start, to =end)
 data(chromhmmNoPattern)
 plotTracks(chromhmmNoPattern, from = start, to =end)
}
```

chromatinHMMOne

Creating one chromHMM track from the UCSC genome browser

## **Description**

Create one track of only one type of chromHMM Broad element from the UCSC genome browser using the Gviz bioconductor package

```
chromatinHMMOne(gen, chr, start, end, mySession, track.name = "Broad ChromHMM", table.name = NULL)
```

6 chromatinHMMOne

## **Arguments**

gen	the name of genome
chr	the chromosome of interest
start	the first position in region of interest (the smallest value)
end	the last position in region of interest (the biggest value)
mySession	the object session from the function browserSession of rtracklayer
track.name	the name of the track( Broad ChromHMM )
table.name	the name of the table from the track

#### Value

An AnnotationTrack object of Gviz

## Author(s)

Tiphaine Martin

## **Examples**

```
library("Gviz")
  gen <- "hg19"
  chr <- "chr2"
  start <- 38290160
  end <- 38303219
if(interactive()) {
  BROWSER.SESSION="UCSC"
  mySession <- browserSession(BROWSER.SESSION)</pre>
  genome(mySession) <- gen</pre>
  {\tt track.name="Broad ChromHMM"}
  tablestrack<-tableNames(ucscTableQuery(mySession, track=track.name))</pre>
  table.name<-tablestrack[1]</pre>
  chromhmmtrackone<-chromatinHMMOne(gen,chr,start,end,mySession,track.name,table.name)</pre>
  plotTracks(chromhmmtrackone, from = start, to =end)
}else {
  data(chromhmmtrackone)
  plotTracks(chromhmmtrackone, from = start, to =end)
```

chrUCSC2ENSEMBL 7

chrUCSC2ENSEMBL	Removing "chr" to the chromosome number from UCSC to transform
	it to ENSEMBL chromosome format

## Description

Removing "chr" at the beginning of the chromosome number

#### Usage

```
chrUCSC2ENSEMBL(chr)
```

#### **Arguments**

chr

the chromosome number in UCSC format

#### Value

the number of chromosome at ENSEMBL format

#### Author(s)

Tiphaine Martin

## **Examples**

```
chr<-"chr7"
chrUCSC2ENSEMBL(chr)</pre>
```

ClinVarCnvTrack

Create one track of the genomic positions of variants from the ClinVar database (CNV only)

## Description

Create one track of the genomic positions of variants from the ClinVar database (CNV only, Variants excluded) using the Gviz bioconductor package

```
ClinVarCnvTrack(gen, chr, start, end, showId = FALSE)
```

8 ClinVarMainTrack

## **Arguments**

gen	the name of the genome
chr	the chromosome of interest
start	the first position in region of interest (the smallest value)
end	the last position in region of interest (the biggest value)

showId Show the ID of the genetic elements

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

### **Examples**

```
library("Gviz")
chrom <- "chr2"
start <- 38290160
end <- 38303219
gen <- "hg19"
if(interactive()){
   clinCNV<-ClinVarCnvTrack(gen,chrom,start,end)
   plotTracks(clinCNV, from = start, to =end)
}else {
   data(ClinVarCnvTrack)
   plotTracks(clinCNV, from = start, to =end)
}</pre>
```

ClinVarMainTrack

Create one track of the genomic positions of variants from the ClinVar database (variants only)

#### **Description**

Create one track of the genomic positions of variants from the ClinVar database (Variants only, CNV excluded) using the Gviz bioconductor package

```
ClinVarMainTrack(gen, chr, start, end, showId=FALSE)
```

## Arguments

gen	the name of the genome
chr	the chromosome of interest
start	the first position in region of interest (the smallest value)
end	the last position in region of interest (the biggest value)
showId	Show the ID of the genetic elements

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chrom <- "chr2"
start <- 38290160
end <- 38303219

if(interactive()) {
    clinVariant<-ClinVarMainTrack(gen,chrom,start,end)
    plotTracks(clinVariant, from = start, to =end)
}else{
    data(clinVarMaintrack)
    plotTracks(clinVariant, from = start, to =end)
}</pre>
```

comet

Visualize EWAS results in a genomic region of interest

#### **Description**

coMET is an R-based package to visualize EWAS (epigenome-wide association scans) results in a genomic region of interest. The main feature of coMET is to plot the the significance level of EWAS results in the selected region, along with correlation in DNA methylation values between CpG sites in the region. The coMET package generates plots of phenotype-association, co-methylation patterns, and a series of annotation tracks.

```
comet(MYDATA.FILE = NULL, MYDATA.FORMAT = "SITE", MYDATA.LARGE.FILE = NULL, MYDATA.LARGE.FORMAT = "SITE"
```

#### **Arguments**

MYDATA.FILE Name of the info file describing the coMET parameters

MYDATA. FORMAT Format of the input data in MYDATA.FILE. There are 4 different options: SITE,

REGION, SITE\_ASSOC, REGION\_ASSOC

MYDATA.LARGE.FILE

Name of additional info files describing the coMET parameters. File names should be comma-separated.

MYDATA.LARGE.FORMAT

Format of additional data to be visualised in the p-value plot. File names should be comma-separated.

CORMATRIX.FILE Name of the raw data file or the pre-computed correlation matrix file.

CORMATRIX.METHOD

Options for calculating the correlation matrix: spearman, pearson and kendall

CORMATRIX.FORMAT

Format of the input CORMATRIX.FILE. TThere are two options: raw file (RAW if CpG sites are by column and samples by row or RAW\_REV if CpG site are by row and samples by column) and pre-computed correlation matrix (CORMATRIX)

CORMATRIX.COLOR.SCHEME

Color scheme options: heat, bluewhitered, cm, topo, gray, bluetored

MYDATA.REF The name of the reference CpG-site listed in MYDATA.FILE

START The first nucleotide position to be visualised END the last nucleotide position to be visualised

ZOOM Default=False

LAB. Y Scale of the y-axis. Options: log or ln

PVAL.THRESHOLD Significance threshold to be displayed as a red dashed line

DISP.PVAL.THRESHOLD

Display only the findings that pass the value put in DISP.PVAL.THRESHOLD

#### DISP.ASSOCIATION

This logical option works only if MYDATA.FILE contains the effect direction (MYDATA.FORMAT=SITE\_ASSOC or REGION\_ASSOC). The value can be TRUE or FALSE: if FALSE (default), for each point of data in the p-value plot, the color of symbol is the color of co-methylation pattern between the point and the reference site; if TRUE, the effect direction is shown. If the association is positive, the color is the one defined with the option COLOR.LIST. On the other hand, if the association is negative, the color is the opposed color.

#### DISP.ASSOCIATION.LARGE

This logical option works only if MYDATA.LARGE.FILE contains the effect direction (MYDATA.LARGE.FORMAT=SITE\_ASSOC or REGION\_ASSOC). The value can be TRUE or FALSE: if FALSE (default), for each point of data in the p-value plot, the color of symbol is the color of co-methylation pattern between the point and the reference site; if TRUE, the effect direction is shown. If the association is positive, the color is the one defined with the option COLOR.LIST.LARGE. On the other hand, if the association is negative, the color is the opposed color.

DISP.REGION

This logical option works only if MYDATA.FILE contains regions (MYDATA.FORMAT=REGION or REGION\_ASSOC). The value can be TRUE or FALSE (default). If TRUE, the genomic element will be shown by a continuous line with the color of the element, in addition to the symbol at the center of the region. If FALSE, only the symbol is shown.

DISP.REGION.LARGE

This logical option works only if MYDATA.LARGE.FILE contains regions (MY-DATA.LARGE.FORMAT=REGION or REGION\_ASSOC). The value can be TRUE or FALSE (default). If TRUE, the genomic element will be shown by a continuous line with the color of the element, in addition to the symbol at the center of the region. If FALSE, only the symbol is shown.

**SYMBOLS** 

The symbol shown in the p-value plot. Options: circle, square, diamond, triangle. Symbols can be filled by appending -fill, e.g. square-fill. Example: circle, diamond-fill, triangle

SYMBOLS.LARGE

The symbol to visualise the data defined in MYDATA.LARGE.FILE. Options: circle, square, diamond, triangle; Symbols can either be filled or not filled by appending -fill e.s., square-fill. Example: circle, diamond-fill, triangle

SAMPLE.LABELS

Labels for the sample described in MYDATA.FILE to include in the legend

SAMPLE.LABELS.LARGE

Labels for the sample described in MYDATA.LARGE.FILE to include in the legend

USE.COLORS

Use the colors defined or use the grey color scheme

DISP. COLOR. REF Logical option TRUE or FALSE (TRUE default). if TRUE, the connection line related to the reference probe is in purple, if FALSE if the connection line related to the reference probe stay black.

COLOR.LIST

List of colors for displaying the P-value symbols related to the data in MY-DATA.FILE

COLOR.LIST.LARGE

List of colors for displaying the P-value symbols related to the data in MY-DATA.LARGE.FILE

DISP.MYDATA

logical option TRUE or FALSE. TRUE (default). If TRUE, the P-value plot is shown; if FALSE the plot will be defined by GViz

BIOFEAT.USER.FILE

Name of data file to visualise in the tracks. File names should be commaseparated.

BIOFEAT.USER.TYPE

Track type, where multiple tracks can be shown (comma-separated): DataTrack, AnnotationTrack, GeneRegionTrack.

BIOFEAT.USER.TYPE.PLOT

Format of the plot if the data are shown with the Gviz's function called Data-Track (comma-separated)

GENOME The human genome reference file. e.g. "hg19" for Human genome 19 (NCBI

DATASET.GENE The gene names from ENSEMBL. e.g. hsapiens\_gene

Name of SNP database from ENSEMBL; Default: hsapiens\_snp DATASET.SNP

VERSION. DBSNP Name of dbSNP used; Default: snp138 version from DBSNP

DATASET.SNP.STOMA

Optional. Name of somatic SNP database from ENSEMBL. Default : hsapiens\_snp\_som

DATASET.REGULATION

Optional. Name of regulation database from ENSEMBL. Default: hsapiens\_feature\_set

DATASET.STRU Optional. Name of structural variation database from ENSEMBL. Default: hsapiens\_structvar

DATASET.STRU.STOMA

Optional. Name of somatic structural variation database from ENSEMBL. Default: hsapiens\_structvar\_som

PATTERN. REGULATION

Name of cell type of DATASET.REGULATION. Default: GM12878

BROWSER.SESSION

Name of database for BioMART connection. Default: UCSC

TRACKS.GVIZ list of tracks created by Gviz.

TRACKS.GGBIO list of tracks created by ggbio.

TRACKS.TRACKVIEWER

list of tracks created by track viewer.

DISP.MYDATA.NAMES

logical option TRUE or FALSE. If True (default), the names of the CpG sites are displayed.

DISP. COLOR. BAR Color legend for the correlation matrix (range -1 to 1). Default: blue-white-red

DISP.PHYS.DIST logical option (TRUE or FALSE). TRUE (default). Display the bp distance on the plots

DISP. LEGEND logical option TRUE or FALSE. TRUE (default) Display the sample labels and corresponding symbols on the lower right side

DISP.MARKER.LINES

logical option TRUE or FALSE. TRUE (default), if FALSE the red line for PVAL.THRESHOLD is not shown

DISP.CORMATRIXMAP

logical optionTRUE or FALSE. TRUE (default), if FALSE correlation matrix is not shown

DISP.PVALUEPLOT

logical option (TRUE or FALSE). TRUE (default), if FALSE the pvalue plot is not shown

DISP.TYPE Default: symbol

DISP.MULT.LAB.X

logical option TRUE or FALSE. FALSE (default). Display evenly spaced X-axis labels; up to 5 labels are shown.

DISP.CONNECTING.LINES

logical option TRUE or FALSE. TRUE (default) displays connecting lines between p-value plot and correlation matrix

PALETTE.FILE File that contains color scheme for the heatmap. Colors are hexidecimal HTML color codes; one color per line; if you do not want to use this option, use the

color defined by the option CORMATRIX.COLOR.SCHEME

IMAGE.TITLE Title of the plot

IMAGE.NAME Path and Name of the plot file without extension

IMAGE. TYPE Options: pdf or eps

IMAGE.SIZE Default: 3.5 inches. Possible sizes: 3.5 or 7
FONT.FACTOR Font size of the sample labels. Range: 0-1

SYMBOL.FACTOR Size of the symbols. Range: 0-1

PRINT.IMAGE Print image in file or not.

CONNECTING.LINES.FACTOR

Length of the connecting lines. Range: 0-2

CONNECTING.LINES.ADJ

Position of the connecting lines horizontally. Negative values shift the connecting lines to the left and positive values shift the lines to the right. Range: (-1;1) option -1 means no connecting lines.

CONNECTING.LINES.VERT.ADJ

Position of the connecting lines vertically. Can be used to vertically adjust the position of the connecting lines in relation to the CpG-site names. Negative value shift the connecting lines down. Range: (-0.5 - 0), option -1 mean the default value related to the plot size (-0.5 for 3.5 plot size; -0.7 for 7.5 plot size)

CONNECTING.LINES.FLEX

Adjusts the spread of the connecting lines. Range: 0-2

config.file Configuration file that contains the values of these options instead of defining

these by command line

VERBOSE logical option TRUE or FALSE. TRUE (default). If TRUE, shows comments.

#### Value

Create a plot

#### Author(s)

Tiphaine Martin

#### **Examples**

```
extdata <- system.file("extdata", package="coMET",mustWork=TRUE)
configfile <- file.path(extdata, "config_cyp1b1_zoom_4comet.txt")
myinfofile <- file.path(extdata, "cyp1b1_infofile.txt")
myexpressfile <- file.path(extdata, "cyp1b1_infofile_exprGene_region.txt")
mycorrelation <- file.path(extdata, "cyp1b1_res37_rawMatrix.txt")

chrom <- "chr2"
start <- 38290160
end <- 38303219</pre>
```

14 comet.web

```
gen <- "hg19"
if(interactive()){
 cat("interactive")
 genetrack <-genesENSEMBL(gen,chrom,start,end,showId=FALSE)</pre>
 snptrack <- snpBiomart(chrom, start, end,</pre>
                        dataset="hsapiens_snp_som", showId=FALSE)
 strutrack <- structureBiomart(chrom, start, end,</pre>
                               strand, dataset="hsapiens_structvar_som")
 clinVariant<-ClinVarMainTrack(gen,chrom,start,end)</pre>
 clinCNV<-ClinVarCnvTrack(gen,chrom,start,end)</pre>
 gwastrack <-GWASTrack(gen,chrom,start,end)</pre>
 geneRtrack <-GeneReviewsTrack(gen,chrom,start,end)</pre>
 listgviz <- list(genetrack,snptrack,strutrack,clinVariant,</pre>
                 clinCNV,gwastrack,geneRtrack)
comet(config.file=configfile, MYDATA.FILE=myinfofile, CORMATRIX.FILE=mycorrelation,
      MYDATA.LARGE.FILE=myexpressfile, TRACKS.GVIZ=listgviz,
      VERBOSE=FALSE, PRINT.IMAGE=FALSE, DISP.PVALUEPLOT=FALSE)
} else {
 cat("Non interactive")
 data(geneENSEMBLtrack)
 data(snpBiomarttrack)
 data(ISCAtrack)
 data(strucBiomarttrack)
 data(ClinVarCnvTrack)
 data(clinVarMaintrack)
 data(GWASTrack)
 data(GeneReviewTrack)
 listgviz <- list(genetrack,snptrack,strutrack,clinVariant,</pre>
                 clinCNV,gwastrack,geneRtrack)
comet(config.file=configfile, MYDATA.FILE=myinfofile, CORMATRIX.FILE=mycorrelation,
      MYDATA.LARGE.FILE=myexpressfile, TRACKS.GVIZ=listgviz,
      VERBOSE=FALSE, PRINT.IMAGE=FALSE, DISP.PVALUEPLOT=FALSE)
}
```

comet.web

Visualize EWAS results in a genomic region of interest with predefined annotation tracks

## **Description**

coMET is an R-based package to visualize EWAS (epigenome-wide association scans) results in a genomic region of interest. The main feature of coMET is to plot the the significance level of EWAS results in the selected region, along with correlation in DNA methylation values between CpG sites in the region. The coMET package generates plots of phenotype-association, co-methylation patterns, and a series of annotation tracks.

comet.web

#### Usage

comet.web(MYDATA.FILE = NULL, MYDATA.FORMAT = c("SITE", "REGION", "SITE\_ASSO", "REGION\_ASSO"), MYDATA.

#### **Arguments**

MYDATA.FILE Name of the info file describing the coMET parameters

MYDATA.FORMAT Format of the input data in MYDATA.FILE. There are 4 different options: SITE,

REGION, SITE\_ASSOC, REGION\_ASSOC

MYDATA.LARGE.FILE

Name of additional info files describing the coMET parameters. File names should be comma-separated.

MYDATA.LARGE.FORMAT

Format of additional data to be visualised in the p-value plot. File names should be comma-separated.

CORMATRIX. FILE Name of the raw data file or the pre-computed correlation matrix file.

CORMATRIX.METHOD

Options for calculating the correlation matrix: spearman, pearson and kendall

CORMATRIX.FORMAT

Format of the input CORMATRIX.FILE. There are three options: raw file (RAW if CpG sites are by column and samples by row or RAW\_REV if CpG site are by row and samples by column) and pre-computed correlation matrix (CORMATRIX)

CORMATRIX.COLOR.SCHEME

Color scheme options: heat, bluewhitered, cm, topo, gray, bluetored

MYDATA.REF The name of the reference CpG-site listed in MYDATA.FILE

START The first nucleotide position to be visualised END the last nucleotide position to be visualised

ZOOM logical option TRUE or FALSE. FALSE (default)

LAB. Y Scale of the y-axis. Options: log or ln

PVAL.THRESHOLD Significance threshold to be displayed as a red dashed line

DISP.PVAL.THRESHOLD

Display only the findings that pass the value put in DISP.PVAL.THRESHOLD

#### DISP.ASSOCIATION

This logical option works only if MYDATA.FILE contains the effect direction (MYDATA.FORMAT=SITE\_ASSOC or REGION\_ASSOC). The value can be TRUE or FALSE: if FALSE (default), for each point of data in the p-value plot, the color of symbol is the color of co-methylation pattern between the point and the reference site; if TRUE, the effect direction is shown. If the association is positive, the color is the one defined with the option COLOR.LIST. On the other hand, if the association is negative, the color is the opposed color.

#### DISP.ASSOCIATION.LARGE

This logical option works only if MYDATA.LARGE.FILE contains the effect direction (MYDATA.LARGE.FORMA=SITE\_ASSOC or REGION\_ASSOC). The value can be TRUE or FALSE: if FALSE (default), for each point of data

16 comet.web

> in the p-value plot, the color of symbol is the color of co-methylation pattern between the point and the reference site; if TRUE, the effect direction is shown. If the association is positive, the color is the one defined with the option COLOR.LIST.LARGE. On the other hand, if the association is negative, the color is the opposed color.

DISP.REGION

This logical option works only if MYDATA.FILE contains regions (MYDATA.FORMAT=REGION or REGION\_ASSOC). The value can be TRUE or FALSE (default). If TRUE, the genomic element will be shown by a continuous line with the color of the element, in addition to the symbol at the center of the region. If FALSE, only the symbol is shown.

#### DISP.REGION.LARGE

This logical option works only if MYDATA.LARGE.FILE contains regions (MY-DATA.LARGE.FORMAT=REGION or REGION\_ASSOC). The value can be TRUE or FALSE (default). If TRUE, the genomic element will be shown by a continuous line with the color of the element, in addition to the symbol at the center of the region. If FALSE, only the symbol is shown.

**SYMBOLS** 

The symbol shown in the p-value plot. Options: circle, square, diamond, triangle. Symbols can be filled by appending -fill, e.g. square-fill. Example: circle, diamond-fill, triangle

SYMBOLS.LARGE

The symbol to visualise the data defined in MYDATA.LARGE.FILE. Options: circle, square, diamond, triangle; Symbols can either be filled or not filled by appending -fill e.s., square-fill. Example: circle,diamond-fill,triangle

SAMPLE.LABELS

Labels for the sample described in MYDATA.FILE to include in the legend

SAMPLE.LABELS.LARGE

Labels for the sample described in MYDATA.LARGE.FILE to include in the legend

USE.COLORS

Use the colors defined or use the grey color scheme

DISP. COLOR. REF Logical option TRUE or FALSE (TRUE default). if TRUE, the connection line related to the reference probe is in purple, if FALSE if the connection line related to the reference probe stay black.

COLOR.LIST

List of colors for displaying the P-value symbols related to the data in MY-DATA.FILE

COLOR.LIST.LARGE

List of colors for displaying the P-value symbols related to the data in MY-DATA.LARGE.FILE

DTSP. MYDATA

logical option TRUE or FALSE. TRUE (default). If TRUE, the P-value plot is shown; if FALSE the plot will be defined by GViz

BIOFEAT.USER.FILE

Name of data file to visualise in the tracks. File names should be commaseparated.

BIOFEAT.USER.TYPE

Track type, where multiple tracks can be shown (comma-separated): DataTrack, AnnotationTrack, GeneRegionTrack.

BIOFEAT.USER.TYPE.PLOT

Format of the plot if the data are shown with the Gviz's function called Data-Track (comma-separated)

CoreillCNVTrack 17

LIST.TRACKS List of annotation tracks to visualise. Options include geneENSEMBL, CGI,

ChromHMM, DNAse, RegENSEMBL, SNP, transcriptENSEMBL, SNPstoma, SNPstru, SNPstrustoma, ISCA, COSMIC, GAD, ClinVar, GeneReviews, GWAS,

ClinVarCNV, GCcontent, genesUCSC, xenogenesUCSC.

PATTERN. REGULATION

The cell/tissue or the list of cells/tissues to visualise in the regulation region

defined by Broad ChromHMM

IMAGE.TITLE Title of the plot

IMAGE.NAME path and Name of the plot file without extension

IMAGE.TYPE Options: pdf or eps

IMAGE.SIZE Default: 3.5 inches. Possible sizes: 3.5 or 7
FONT.FACTOR Font size of the sample labels. Range: 0-1

SYMBOL.FACTOR Size of the symbols. Range: 0-1

PRINT.IMAGE Print image in file or not.

config.file Configuration file that contains the values of these options instead of defining

these by command line

VERBOSE logical option TRUE or FALSE. TRUE (default). If TRUE, shows comments.

#### Author(s)

Tiphaine Martin

## **Examples**

CoreillCNVTrack	Create one track of the genomic positions of CNV in chromosomal
	aberration and inherited disorders from the NIGMS Human Genetic
	Cell Repository data

#### **Description**

Create one track of the genomic positions of copy-number variants (CNVs) in chromosomal aberration and inherited disorder cell lines from the NIGMS Human Genetic Cell Repository using the Gviz bioconductor package

18 COSMICTrack

#### Usage

```
CoreillCNVTrack(gen, chr, start, end, showId=FALSE)
```

#### **Arguments**

gen the name of the genome chr the chromosome of interest

start the first position in the region of interest (the smallest value) end the last position in the region of interest (the largest value)

showId Show the ID of the genetic elements

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

## Examples

```
library("Gviz")
gen <- "hg19"
chrom <- "chr2"
start <- 38290160
end <- 38303219

if(interactive()){
   coreilVariant<-CoreillCNVTrack(gen,chrom,start,end)
   plotTracks(coreilVariant, from = start, to =end)
} else {
   data(coreilVarianttrack)
   plotTracks(coreilVariant, from = start, to =end)
}</pre>
```

COSMICTrack

Create one track of the genomic positions of variants from COSMIC

#### **Description**

Create one track of the genomic positions of variants from COSMIC, the "Catalogue Of Somatic Mutations In Cancer" using the Gviz bioconductor package

```
COSMICTrack(gen, chr, start, end, showId=FALSE)
```

cpgIslandsUCSC 19

## **Arguments**

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
showId	Show the ID of the genetic elements

## Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
chrom <- "chr2"
start <- 38290160
end <- 38303219
gen <- "hg19"
if(interactive()){
  cosmicVariant<-COSMICTrack(gen,chrom,start,end)
  plotTracks(cosmicVariant, from = start, to =end)
}else {
  data(cosmicVarianttrack)
  plotTracks(cosmicVariant, from = start, to =end)
}</pre>
```

cpgIslandsUCSC

create track CpG Island from UCSC

#### **Description**

create track CpG Island from UCSC using the Gviz bioconductor package

## Usage

```
cpgIslandsUCSC(gen, chr, start, end)
```

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)

20 DNAseUCSC

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
chrom <- "chr2"
start <- 38290160
end <- 38303219
gen <- "hg19"

if(interactive()) {
  cpgIstrack<-cpgIslandsUCSC(gen, chrom, start, end)
  plotTracks(cpgIstrack, from = start, to =end)
}else {
  data(cpgIslandtrack)
  plotTracks(cpgIstrack, from = start, to =end)
}</pre>
```

DNAseUCSC

Creation of an UCSC's DNase clusters track

### **Description**

Creation of DNase cluster track from a connection to UCSC genome browser in using the GViz bioconductor package

## Usage

```
DNAseUCSC(gen, chr, start, end, mySession, track.name = "DNase Clusters", table.name = NULL)
```

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
mySession	the object session from the function browserSession of rtracklayer
track.name	the name of the track DNAseUCSC. "DNase Clusters"(default)
table.name	the name of the table from the track

GADTrack 21

#### Value

An AnnotationTrack object of Gviz

#### Author(s)

Tiphaine Martin

## **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219
if(interactive()){
  BROWSER.SESSION="UCSC"
  mySession <- browserSession(BROWSER.SESSION)</pre>
  genome(mySession) <- gen</pre>
  track.name="Broad ChromHMM"
  tablestrack<-tableNames(ucscTableQuery(mySession, track=track.name))</pre>
  table.name<-tablestrack[1]
  dnasetrack<-DNAseUCSC(gen,chr,start,end,mySession)</pre>
  plotTracks(dnasetrack, from = start, to =end)
}else {
    data(dnasetrack)
   plotTracks(dnasetrack, from = start, to =end)
}
```

GADTrack

Create one track of the genomic positions of variants from the Genetic Association Database (GAD)

## Description

Create one track of the genomic positions of variants from the Genetic Association Database (GAD) (archive of human genetic association studies of complex diseases and disorders) using the Gviz bioconductor package

#### Usage

```
GADTrack(gen, chr, start, end, showId=FALSE)
```

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)

22 gcContent

end the last position in the region of interest (the largest value)

showId Show the ID of the genetic elements

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

## **Examples**

```
library("Gviz")
gen2 <- "hg19"
chrom2 <- "chr2"
start2 <- 38290160
end2 <- 38303219

if(interactive()) {
   gadtrack<-GADTrack(gen=gen2 ,chr=chrom2 ,start=start2 ,end=end2)
   plotTracks(gadtrack, from = start2, to =end2)
} else {
   data(gadtrack)
   plotTracks(gadtrack, from = start2, to =end2)
}</pre>
```

gcContent

Create one track of GC content from UCSC

#### **Description**

Create a track of GC content from UCSC using the Gviz bioconductor package

#### Usage

```
gcContent(gen, chr, start, end)
```

## Arguments

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)

end the last position in the region of interest (the largest value)

GeneReviewsTrack 23

#### Value

A UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219

if(interactive()){
   gctrack<-gcContent(gen,chr,start,end)
   plotTracks(gctrack,from= start, to=end)
} else {
   data(gctrack)
   plotTracks(gctrack,from= start, to=end)
}</pre>
```

GeneReviewsTrack

Create one track of the genomic positions of variants from GeneReviews

## Description

Create one track of the genomic positions of variants from GeneReviews using the Gviz bioconductor package

#### Usage

```
GeneReviewsTrack(gen, chr, start, end, showId=FALSE)
```

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
showId	Show the ID of the genetic elements

24 genesENSEMBL

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

## **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr2"
start <- 38290160
end <- 38303219
if(interactive()){
   geneRtrack <-GeneReviewsTrack(gen,chrom,start,end)
   plotTracks(geneRtrack, from = start, to = end)
} else {
   data(GeneReviewTrack)
   plotTracks(geneRtrack, from = start, to = end)
}</pre>
```

genesENSEMBL

Create one track of the genes in the genomic regions of interest from EMSEMBL

## Description

Create one track of the genes in the genomic regions of interest from EMSEMBL using the Gviz bioconductor package

#### **Usage**

```
genesENSEMBL(gen, chr, start, end, showId=FALSE)
```

## Arguments

gen the name of the genome chr the chromosome of interest

start the first position in the region of interest (the smallest value) end the last position in the region of interest (the largest value)

showId Show the ID of the genetic elements

#### Value

A BiomartGeneRegionTrack object of Gviz

genesNameENSEMBL 25

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr2"
start <- 38290160
end <- 38303219
if(interactive()) {
  genetrack <-genesENSEMBL(gen,chrom,start,end,showId=FALSE)
  plotTracks(genetrack, from = start, to =end)
} else {
  data(geneENSEMBLtrack)
  plotTracks(genetrack, from = start, to =end)
}</pre>
```

genesNameENSEMBL

Obtain the genes names in the genomic regions of interest from EN-SEMBL

## Description

Obtain the genes names in the genomic regions of interest from ENSEMBL

## Usage

```
genesNameENSEMBL(gen, chr, start, end, dataset)
```

## **Arguments**

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
dataset	Name of the database to select genes

## Author(s)

26 GWASTrack

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219

if(interactive()){
   dataset<- "hsapiens_gene_ensemb1"
   geneNameEnsemb1<- genesNameENSEMBL(gen,chr,start,end,dataset)
   geneNameEnsemb1
} else {
   data(geneNameEnsemb1)
   geneNameEnsemb1
}</pre>
```

GWASTrack

Create one track of the genomic positions of variants from the GWAS catalog

## Description

Create one track of the genomic positions of variants from the NHGRI Catalog of Published Genome-Wide Association Studies using the Gviz bioconductor package

#### Usage

```
GWASTrack(gen, chr, start, end, showId=FALSE)
```

## Arguments

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (

start the first position in the region of interest (the smallest value) end the last position in the region of interest (the largest value)

showId Show the ID of the genetic elements

## Value

An UcscTrack object of Gviz

## Author(s)

HistoneAll 27

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr2"
start <- 37949607
end <- 37965207

if(interactive()) {
   gwastrack <-GWASTrack(gen,chrom,start,end)
   plotTracks(gwastrack, from = start, to =end)
} else {
   data(GWASTrack)
   plotTracks(gwastrack, from = start, to =end)
}</pre>
```

HistoneAll

Create multiple tracks of histone modifications from the UCSC genome browser

## Description

Create multiple tracks of histone modifications from the UCSC genome browser (ENCODE/Broad) using the Gviz bioconductor package

#### Usage

```
HistoneAll(gen, chr, start, end, mySession, pattern = NULL, track.name = "Broad Histone", table.name = N
```

## **Arguments**

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
mySession	the object session from the function browserSession of rtracklayer
pattern	The cell type
track.name	the name of the track, for example: "Broad Histone"
table.name	the name of the table from the track

## Value

A list of AnnotationTrack object of Gviz

#### Author(s)

28 HistoneOne

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr2"
start <- 38290160
end <- 38313219
if(interactive()){
  BROWSER.SESSION="UCSC"
  mySession <- browserSession(BROWSER.SESSION)</pre>
  genome(mySession) <- gen</pre>
  pattern1 <- "GM12878"
 histonalltrack<-HistoneAll(gen,chr,start,end,mySession, pattern=pattern1,track.name="Broad Histone")
  plotTracks(histonalltrack, from = start, to =end)
} else {
  data(histonalltrack)
  plotTracks(histonalltrack, from = start, to =end)
}
```

HistoneOne

Create one track of one histone modification profile from the UCSC genome browser

## Description

Create one track of one histone modification profile from the UCSC genome browser (ENCODE/Broad) using the Gviz bioconductor package

## Usage

```
HistoneOne(gen, chr, start, end, mySession, track.name = "Broad Histone", table.name = NULL)
```

## Arguments

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
mySession	the object session from the function browserSession of rtracklayer
track.name	the name of the track, for example: "Broad Histone"
table.name	the name of the table from the track

#### Value

An AnnotationTrack object of Gviz

ISCATrack 29

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr2"
start <- 38290160
end <- 38303219

if(interactive()) {
   BROWSER.SESSION="UCSC"
   mySession <- browserSession(BROWSER.SESSION)
   genome(mySession) <- gen
   histoneonetrack<-HistoneOne(gen,chr,start,end,mySession)
   plotTracks(histoneonetrack, from = start, to =end)
} else {
   data(histoneonetrack)
   plotTracks(histoneonetrack, from = start, to =end)
}</pre>
```

**ISCATrack** 

Create one track of the genomic positions of variants from ISCA

#### **Description**

Create one track of the genomic positions of variants from International Standards for Cytogenomic Arrays (ISCA) Consortium using the Gviz bioconductor package

## Usage

```
ISCATrack(gen, chr, start, end, mySession, table.name, showId=FALSE)
```

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
mySession	the object session from the function browserSession of rtracklayer
table.name	A table of ISCAT classifications: iscaBenign, iscaCuratedBenign, iscaCuratedPathogenic, iscaLikelyBenign, iscaLikelyPathogenic, iscaPathGainCum, iscaPathLossCum, iscaPathogenic, iscaUncertain
showId	Show the ID of the genetic elements

30 knownGenesUCSC

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

## **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr2"
start <- 38292433
end <- 38305492

if(interactive()){
    BROWSER.SESSION="UCSC"
    mySession <- browserSession(BROWSER.SESSION)
    genome(mySession) <- gen
    iscatrack <-ISCATrack(gen,chrom,start,end,mySession, table="iscaPathogenic")
    plotTracks(iscatrack, from = start, to =end)
} else {
    data(ISCAtrack)
    plotTracks(iscatrack, from = start, to =end)
}</pre>
```

knownGenesUCSC

Create a track of known genes from the UCSC genome browser

#### **Description**

Create a track of known genes from the UCSC genome browser using the Gviz bioconductor package

## Usage

```
knownGenesUCSC(gen, chr, start, end)
```

# **Arguments** gen

J	$\varepsilon$
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)

the name of the genome

regulationBiomart 31

#### Value

An UcscTrack object of Gviz

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219

if(interactive()) {
   genesUcsctrack<-knownGenesUCSC(gen,chr,start,end)
   plotTracks(genesUcsctrack, from = start, to =end)
}else {
   data(genesUcsctrack)
   plotTracks(genesUcsctrack, from = start, to =end)
}</pre>
```

 ${\tt regulationBiomart}$ 

Create a regulation track from ENSEMBL

#### **Description**

Create a 'Regulation' track from ENSEMBL using the Gviz bioconductor package

## Usage

```
regulationBiomart(chr, start, end, dataset="hsapiens_feature_set")
```

#### **Arguments**

chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
dataset	The name of the database. "hsapiens_feature_set" (default)

## Value

An AnnotationTrack object of Gviz

32 snpBiomart

#### Author(s)

Tiphaine Martin

#### **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219

if(interactive()){
   regulationENSEMBLtrack<-regulationBiomart(chr,start,end,dataset="hsapiens_feature_set")
   plotTracks(regulationENSEMBLtrack, from = start, to =end)
} else {
   data(regulationENSEMBLtrack)
   plotTracks(regulationENSEMBLtrack, from = start, to =end)
}</pre>
```

snpBiomart

Create a short variation track from ENSEMBL

#### **Description**

Create a 'Short Variation' track from ENSEMBL using the Gviz bioconductor package

#### Usage

```
snpBiomart(chr, start, end, dataset, showId=FALSE, title = NULL)
```

## **Arguments**

chr the chromosome of interest
start the first position in the region of interest (the smallest value)
end the last position in the region of interest (the largest value)
dataset The name of the database. Example "hsapiens\_snp\_som"
showId Show the ID of element or not

title The name of the annotation track

#### Value

An AnnotationTrack object of Gviz

#### Author(s)

snpLocationsUCSC 33

#### **Examples**

snpLocationsUCSC

Create a SNP track from UCSC

## Description

Create a SNP track from UCSC using the Gviz bioconductor package

## Usage

```
snpLocationsUCSC(gen, chr, start, end, track)
```

#### **Arguments**

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
track	The name of the database. Example "snp138"

#### Value

An UcscTrack object of Gviz

## Author(s)

34 structureBiomart

## **Examples**

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219

if(interactive()) {
    snpUCSCtrack<-snpLocationsUCSC(gen,chr,start,end,"snp138")
    plotTracks(snpUCSCtrack, from = start, to =end)
} else {
    data(snpUCSCtrack)
    plotTracks(snpUCSCtrack, from = start, to =end)
}</pre>
```

structureBiomart

Create a structural variation track from ENSEMBL

#### **Description**

Create a 'Structural Variation' track from ENSEMBL using the Gviz bioconductor package

## Usage

```
structureBiomart(chr, start, end, strand, dataset, showId=FALSE, title = NULL)
```

## **Arguments**

chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
strand	the strand to extract structure data for
dataset	The name of the database. Example "hsapiens_structvar_som"
showId	Show the ID of the element
title	The name of the annotation track

#### Value

An AnnotationTrack object of Gviz

## Author(s)

transcriptENSEMBL 35

#### **Examples**

transcriptENSEMBL

Create a track of transcripts from ENSEMBL

## Description

Create a track to visualize different transcripts from ENSEMBL using the Gviz bioconductor package

#### Usage

```
transcriptENSEMBL(gen, chr, start, end, showId = FALSE)
```

## Arguments

gen	the name of the genome
chr	the chromosome of interest
start	the first position in the region of interest (the smallest value)
end	the last position in the region of interest (the largest value)
showId	Show the ID of the genetic elements

## Value

A BiomartGeneRegionTrack object of Gviz

## Author(s)

36 transcriptENSEMBL

## Examples

```
library("Gviz")
gen <- "hg19"
chr <- "chr7"
start <- 38290160
end <- 38303219

if(interactive()){
   transENSMBLtrack<-transcriptENSEMBL(gen,chr,start,end)
   plotTracks(transENSMBLtrack, from = start, to =end)
} else {
   data(transENSMBLtrack)
   plotTracks(transENSMBLtrack, from = start, to =end)
}</pre>
```

# **Index**

*Topic <b>dplot</b>	comet.web, 14
chrUCSC2ENSEMBL, 7	CoreillCNVTrack, 17
*Topic <b>hplot</b>	COSMICTrack, 18
chromatinHMMAll,4	cpgIslandsUCSC, 19
chromatinHMMOne, 5	
ClinVarCnvTrack, 7	DNAseUCSC, 20
ClinVarMainTrack,8	
comet, 9	GADTrack, 21
comet.web, 14	gcContent, 22
CoreillCNVTrack, 17	GeneReviewsTrack, 23
COSMICTrack, 18	genesENSEMBL, 24
cpgIslandsUCSC, 19	genesNameENSEMBL, 25
DNAseUCSC, 20	GWASTrack, 26
GADTrack, 21	HistoneAll, 27
gcContent, 22	HistoneOne, 28
GeneReviewsTrack, 23	nistolleone, 26
genesENSEMBL, 24	ISCATrack, 29
GWASTrack, 26	2007.11 001., 22
HistoneAll, 27	knownGenesUCSC, 30
HistoneOne, 28	
ISCATrack, 29	regulationBiomart, $31$
knownGenesUCSC, 30	
regulationBiomart,31	snpBiomart, 32
snpBiomart, 32	snpLocationsUCSC, 33
snpLocationsUCSC, 33	structureBiomart, 34
structureBiomart, 34	transprintENCEMPL 25
transcriptENSEMBL, 35	transcriptENSEMBL, 35
*Topic <b>misc</b>	
genesNameENSEMBL, 25	
*Topic <b>package</b>	
coMET-package, 2	
chromatinHMMAll, 4	
chromatinHMMOne, 5	
chrUCSC2ENSEMBL, 7	
ClinVarCnvTrack, 7	
ClinVarMainTrack, 8	
coMET (coMET-package), 2	
comet, 9	
coMET-package, 2	