

Package ‘GenVisR’

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Title Genomic Visualizations in R

Version 1.18.1

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Description Produce highly customizable publication quality graphics for genomic data primarily at the cohort level.

Depends R (>= 3.3.0), methods

Imports AnnotationDbi, biomaRt, BiocGenerics, Biostrings, DBI, FField, GenomicFeatures, GenomicRanges (>= 1.25.4), ggplot2 (>= 2.1.0), grid, gridExtra (>= 2.0.0), gtable, gtools, IRanges (>= 2.7.5), plyr (>= 1.8.3), reshape2, Rsamtools, scales, stats, utils, viridis, data.table, BSgenome, grDevices, GenomeInfoDb, VariantAnnotation

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BugReports <https://github.com/griffithlab/GenVisR/issues>

biocViews Infrastructure, DataRepresentation, Classification, DNASEq

LazyData true

Suggests BiocStyle, BSgenome.Hsapiens.UCSC.hg19, knitr, RMySQL, roxygen2, testthat, TxDb.Hsapiens.UCSC.hg19.knownGene, rmarkdown, vdiff, formatR, TxDb.Hsapiens.UCSC.hg38.knownGene, BSgenome.Hsapiens.UCSC.hg38

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Collate 'AllGenerics.R' 'Clinical-class.R' 'GMS_Virtual-class.R' 'GMS-class.R' 'GMS_v4-class.R' 'GenVisR.R' 'Lollipop-class.R' 'MutSpectra-class.R' 'MutationAnnotationFormat_Virtual-class.R' 'MutationAnnotationFormat-class.R' 'MutationAnnotationFormat_v1.0-class.R' 'MutationAnnotationFormat_v2.0-class.R' 'MutationAnnotationFormat_v2.1-class.R' 'MutationAnnotationFormat_v2.2-class.R' 'MutationAnnotationFormat_v2.3-class.R'

'MutationAnnotationFormat_v2.4-class.R' 'Rainfall-class.R'
 'VEP_Virtual-class.R' 'VEP-class.R' 'VEP_v88-class.R'
 'Waterfall-class.R' 'cnFreq.R' 'cnFreq_buildMain.R'
 'cnFreq_disjoin.R' 'cnFreq_qual.R' 'cnSpec.R'
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 'genCov.R' 'genCov_alignPlot.R'
 'genCov_assign_ggplotGrob_height.R'
 'genCov_assign_ggplotGrob_width.R' 'genCov_buildCov.R'
 'genCov_buildTrack.R' 'genCov_extr_ggplotGrob_height.R'
 'genCov_extr_ggplotGrob_width.R' 'genCov_qual.R'
 'genCov_trackViz.R' 'geneViz.R' 'geneViz_Granges2dataframe.R'
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 'geneViz_mapCoordSpace.R' 'geneViz_mapCovCoordSpace.R'
 'geneViz_mergeRegions.R' 'geneViz_mergeTypeRegions.R'
 'geneViz_mergeTypes.R' 'ideoView.R' 'ideoView_buildMain.R'
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 'lohSpec_tileCalc.R' 'lohSpec_tilePosition.R'
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 'lohView_buildMain.R' 'lohView_qual.R' 'multi_align.R'
 'multi_buildClin.R' 'multi_chrBound.R' 'multi_cytobandRet.R'
 'multi_selectOut.R' 'multi_subsetChr.R'

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|---------|--------------------------------|
| brcaMAF | <i>Truncated BRCA MAF file</i> |
|---------|--------------------------------|

Description

A data set containing 50 samples corresponding to "Breast invasive carcinoma" originating from the TCGA project in .maf format (version 2.4): <https://wiki.nci.nih.gov/display/TCGA/TCGA+MAF+Files#TCGAMAFFile>
 BRCA:Breastinvasivecarcinoma, /dccfiles_prod/tcgafiles/distro_ftpusers/anonymous/tumor/brca/gsc/genome.wustl.edu/

Usage

```
data(brcaMAF)
```

Format

a data frame with 2773 observations and 55 variables

Value

Object of class data drame

| | |
|----------------|-----------------------|
| Clinical-class | <i>Class Clinical</i> |
|----------------|-----------------------|

Description

An S4 class to store clinical information and plots, under development!!!

Usage

```
Clinical(path, inputData = NULL, inputFormat = c("wide", "long"),
  legendColumns = 1, palette = NULL, clinicalLayers = NULL,
  verbose = FALSE)
```

Arguments

| | |
|----------------|---|
| path | String specifying the path to clinical data, file must have the column "sample". |
| inputData | Optional data.table or data.frame object holding clinical data, used only if path is not specified. Data must have the column "sample". |
| inputFormat | String specifying the input format of the data given, one of wide or long format (see details). |
| legendColumns | Integer specifying the number of columns in the legend. |
| palette | Named character vector supplying colors for clinical variables. |
| clinicalLayers | list of ggplot2 layers to be passed to the plot. |
| verbose | Boolean specifying if progress should be reported. |

Details

The `Clinical()` function is a constructor to create a `GenVisR` object of class `Clinical`. This is used to both display clinical data in the form of a heatmap and to add clinical data to various `GenVisR` plots. Input to this function can be either the path to a file containing clinical information using the parameter `"path"`, or alternatively a `data.table` object if this information into R. By default the input is assumed to be in a wide format where each variable has it's own column, in such cases the data will be coerced into a long format where there is a key->value pair mapping to the data. The assumption of "wide"/"long" format can be changed with the `"inputFormat"` parameter, in both cases there should be a column called `"sample"` within the data supplied which is used as an id variable.

Slots

`clinicalGrob` `gtable` object for the clinical plot.

`clinicalLayers` list of `ggtheme` or `ggproto` objects used to build the plot.

`clinicalData` `data.table` object to store clinical data

See Also

[getData](#)

[drawPlot](#)

cnFreq

Construct copy-number frequency plot

Description

Given a data frame construct a plot to display copy number changes across the genome for a group of samples.

Usage

```
cnFreq(x, CN_low_cutoff = 1.5, CN_high_cutoff = 2.5,
       plot_title = NULL, CN_Loss_colour = "#002EB8",
       CN_Gain_colour = "#A30000", x_title_size = 12, y_title_size = 12,
       facet_lab_size = 10, plotLayer = NULL, plotType = "proportion",
       genome = "hg19", plotChr = NULL, out = "plot")
```

Arguments

| | |
|-----------------------------|---|
| <code>x</code> | Object of class data frame with rows representing genomic segments. The data frame must contain columns with the following names <code>"chromosome"</code> , <code>"start"</code> , <code>"end"</code> , <code>"segmean"</code> , and <code>"sample"</code> . |
| <code>CN_low_cutoff</code> | Numeric value representing the point at or below which copy number alterations are considered losses. Only used if <code>x</code> represents CN values. |
| <code>CN_high_cutoff</code> | Numeric value representing the point at or above which copy number alterations are considered gains. Only used if <code>x</code> represents CN values. |
| <code>plot_title</code> | Character string specifying the title to display on the plot. |
| <code>CN_Loss_colour</code> | Character string specifying the colour value for copy number losses. |

| | |
|----------------|---|
| CN_Gain_colour | Character string specifying the colour value for copy number gains. |
| x_title_size | Integer specifying the size of the x-axis title. |
| y_title_size | Integer specifying the size of the y-axis title. |
| facet_lab_size | Integer specifying the size of the faceted labels plotted. |
| plotLayer | Valid ggplot2 layer to be added to the plot. |
| plotType | Character string specifying the type of values to plot. One of "proportion" or "frequency" |
| genome | Character string specifying a valid UCSC genome (see details). |
| plotChr | Character vector specifying specific chromosomes to plot, if NULL all chromosomes for the genome selected are displayed. |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |

Details

cnFreq requires the location of chromosome boundaries for a given genome assembly in order to ensure the entire chromosome space is plotted. As a convenience this information is available to cnSpec for the following genomes "hg19", "hg38", "mm9", "mm10", "rn5" and can be retrieved by supplying one of the afore mentioned assemblies via the 'genome' parameter. If a genome assembly is supplied to the 'genome' parameter and is unrecognized cnSpec will attempt to query the UCSC MySQL database for the required information. If genomic segments are not identical across all samples the algorithm will attempt to perform a disjoint operation splitting existing segments such that there are no overlaps. The 'plotLayer' parameter can be used to add an additional layer to the ggplot2 graphic (see vignette).

Value

One of the following, a dataframe containing data to be plotted, a grob object, or a plot.

Examples

```
# plot on internal GenVisR dataset
cnFreq(LucCNseg)
```

| | |
|--------|--|
| cnSpec | <i>Construct copy-number cohort plot</i> |
|--------|--|

Description

Given a data frame construct a plot to display copy-number calls for a cohort of samples.

Usage

```
cnSpec(x, y = NULL, genome = "hg19", plot_title = NULL,
       CN_Loss_colour = "#002EB8", CN_Gain_colour = "#A30000",
       x_title_size = 12, y_title_size = 12, facet_lab_size = 10,
       plotLayer = NULL, out = "plot", CNscale = "absolute")
```

Arguments

| | |
|----------------|---|
| x | Object of class data frame with rows representing copy-number segment calls. The data frame must contain columns with the following names "chromosome", "start", "end", "segmean", "sample". |
| y | Object of class data frame with rows representing chromosome boundaries for a genome assembly. The data frame must contain columns with the following names "chromosome", "start", "end" (optional: see details). |
| genome | Character string specifying a valid UCSC genome (see details). |
| plot_title | Character string specifying title to display on the plot. |
| CN_Loss_colour | Character string specifying the colour value of copy number losses. |
| CN_Gain_colour | Character string specifying the colour value of copy number gains. |
| x_title_size | Integer specifying the size of the x-axis title. |
| y_title_size | Integer specifying the size of the y-axis title. |
| facet_lab_size | Integer specifying the size of the faceted labels plotted. |
| plotLayer | Valid ggplot2 layer to be added to the plot. |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |
| CNscale | Character string specifying if copy number calls supplied are relative (i.e. copy neutral == 0) or absolute (i.e. copy neutral ==2). One of "relative" or "absolute" |

Details

cnSpec requires the location of chromosome boundaries for a given genome assembly in order to ensure the entire chromosome space is plotted. As a convenience this information is available to cnSpec for the following genomes "hg19", "hg38", "mm9", "mm10", "rn5" and can be retrieved by supplying one of the afore mentioned assemblies via the 'genome' parameter. If a genome assembly is supplied to the 'genome' parameter and is unrecognized cnSpec will attempt to query the UCSC MySQL database for the required information. If chromosome boundary locations are unavailable for a given assembly or if it is desireable to plot a specific region encapsulating the copy number data these boundaries can be supplied to the 'y' paramter which has priority of the parameter 'genome'.

The 'plotLayer' parameter can be used to add an additional layer to the ggplot2 graphic (see vignette).

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
cnSpec(LucCNseg, genome="hg19")
```

cnView

*Construct copy-number single sample plot***Description**

Given a data frame construct a plot to display raw copy number calls for a single sample.

Usage

```
cnView(x, y = NULL, z = NULL, genome = "hg19", chr = "chr1",
       CNscale = "absolute", ideogram_txtAngle = 45, ideogram_txtSize = 5,
       plotLayer = NULL, ideogramLayer = NULL, out = "plot",
       segmentColor = NULL)
```

Arguments

| | |
|-------------------|---|
| x | Object of class data frame with rows representing copy number calls from a single sample. The data frame must contain columns with the following names "chromosome", "coordinate", "cn", and optionally "p_value" (see details). |
| y | Object of class data frame with rows representing cytogenetic bands for a chromosome. The data frame must contain columns with the following names "chrom", "chromStart", "chromEnd", "name", "gieStain" for plotting the ideogram (optional: see details). |
| z | Object of class data frame with row representing copy number segment calls. The data frame must contain columns with the following names "chromosome", "start", "end", "segmean" (optional: see details) |
| genome | Character string specifying a valid UCSC genome (see details). |
| chr | Character string specifying which chromosome to plot one of "chr.." or "all" |
| CNscale | Character string specifying if copy number calls supplied are relative (i.e. copy neutral == 0) or absolute (i.e. copy neutral ==2). One of "relative" or "absolute" |
| ideogram_txtAngle | Integer specifying the angle of cytogenetic labels on the ideogram subplot. |
| ideogram_txtSize | Integer specifying the size of cytogenetic labels on the ideogram subplot. |
| plotLayer | Valid ggplot2 layer to be added to the copy number plot. |
| ideogramLayer | Valid ggplot2 layer to be added to the ideogram sub-plot. |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |
| segmentColor | Character string specifying the color of segment lines. Used only if Z is not null. |

Details

cnView is able to plot in two modes specified via the 'chr' parameter, these modes are single chromosome view in which an ideogram is displayed and genome view where chromosomes are faceted. For the single chromosome view cytogenetic band information is required giving the coordinate, stain, and name of each band. As a convenience cnView stores this information for the following genomes "hg19", "hg38", "mm9", "mm10", and "rn5". If the genome assembly supplied to the 'genome' parameter is not one of the 5 afore mentioned genome assemblies cnView will attempt

to query the UCSC MySQL database to retrieve this information. Alternatively the user can manually supply this information as a data frame to the 'y' parameter, input to the 'y' parameter take precedence of input to 'genome'.

cnView is also able to represent p-values for copy-number calls if they are supplied via the "p_value" column in the argument supplied to x. The presence of this column in x will set a transparency value to copy-number calls with calls of less significance becoming more transparent.

If it is available cnView can plot copy-number segment calls on top of raw calls supplied to parameter 'x' via the parameter 'z'.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Create data
chromosome <- 'chr14'
coordinate <- sort(sample(0:106455000, size=2000, replace=FALSE))
cn <- c(rnorm(300, mean=3, sd=.2), rnorm(700, mean=2, sd=.2), rnorm(1000, mean=3, sd=.2))
data <- as.data.frame(cbind(chromosome, coordinate, cn))

# Plot raw copy number calls
cnView(data, chr='chr14', genome='hg19', ideogram_txtSize=4)
```

compIdent

Construct identity snp comparison plot

Description

Given the bam file path, count the number of reads at the 24 SNP locations

Usage

```
compIdent(x, genome, target = NULL, debug = FALSE, mainLayer = NULL,
          covLayer = NULL, out = "plot")
```

Arguments

| | |
|-----------|---|
| x | data frame with rows representing samples and column names "sample_name", "bamfile". Columns should correspond to a sample name and a bam file path. |
| genome | Object of class BSgenome specifying the genome. |
| target | Object of class data frame containing target locations in 1-base format and containing columns names "chr", "start", "end", "var", "name". Columns should correspond to chromosome, start, end, variant allele, name of location. |
| debug | Boolean specifying if test datasets should be used for debugging. |
| mainLayer | Valid ggplot2 layer for altering the main plot. |
| covLayer | Valid ggplot2 layer for altering the coverage plot. |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |

Details

compIdent is a function designed to compare samples via variant allele frequencies (VAF) at specific sites. By default these sites correspond to 24 identity snps originating from the hg19 assembly however the user can specify alternate sites via the target parameter. To view the 24 identity snp locations use `GenVisR::SNPloci`.

Samples from the same origin are expected to have similar VAF values however results can skew based on copy number alterations (CNA). The user is expected to ensure no CNA occur at the 24 identity snp sites.

For display and debugging purposes a debug parameter is available which will use predefined data instead of reading in bam files. Note that data in the debug parameter is only available at the aforementioned 24 sites.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Read in BSgenome object (hg19)
library(BSgenome.Hsapiens.UCSC.hg19)
hg19 <- BSgenome.Hsapiens.UCSC.hg19

# Generate plot
compIdent(genome=hg19, debug=TRUE)
```

covBars

Construct an overall coverage cohort plot

Description

Given a matrix construct a plot to display sequencing depth achieved as percentage bars for a cohort of samples.

Usage

```
covBars(x, colour = NULL, plot_title = NULL, x_title_size = 12,
        y_title_size = 12, facet_lab_size = 10, plotLayer = NULL,
        out = "plot")
```

Arguments

| | |
|-----------------------------|---|
| <code>x</code> | Object of class matrix with rows representing the sequencing depth (i.e. number of reads) and columns corresponding to each sample in the cohort and elements of the matrix |
| <code>colour</code> | Character vector specifying colours to represent sequencing depth. |
| <code>plot_title</code> | Character string specifying the title to display on the plot. |
| <code>x_title_size</code> | Integer specifying the size of the x-axis title. |
| <code>y_title_size</code> | Integer specifying the size of the y-axis title. |
| <code>facet_lab_size</code> | Integer specifying the size of the faceted labels plotted. |

`plotLayer` Valid ggplot2 layer to be added to the plot.

`out` Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns).

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Create data
x <- matrix(sample(100000,500), nrow=50, ncol=10, dimnames=list(0:49,paste0("Sample",1:10)))

# Call plot function
covBars(x)
```

| | |
|----------|------------------------------------|
| cytoGeno | <i>Cytogenetic banding dataset</i> |
|----------|------------------------------------|

Description

A data set containing cytogenetic band information for all chromosomes in the following genomes "hg38", "hg19", "mm10", "mm9", "rn5", obtained from the UCSC sql database at genome-mysql.cse.ucsc.edu.

Usage

```
data(cytoGeno)
```

Format

a data frame with 3207 observations and 6 variables

Value

Object of class data frame

| | |
|----------|------------------------|
| drawPlot | <i>Method drawPlot</i> |
|----------|------------------------|

Description

Method drawPlot

Usage

```
drawPlot(object, ...)

## S4 method for signature 'Clinical'
drawPlot(object, ...)

## S4 method for signature 'Lollipop'
drawPlot(object, ...)

## S4 method for signature 'MutSpectra'
drawPlot(object, ...)

## S4 method for signature 'Rainfall'
drawPlot(object, ...)

## S4 method for signature 'Waterfall'
drawPlot(object, ...)
```

Arguments

| | |
|--------|--|
| object | Object of class Waterfall, MutSpectra, or Clinical |
| ... | additional arguments to passed |

Details

The drawPlot method is used to draw plots created by GenVisR plot constructor functions.

| | |
|--------|---|
| genCov | <i>Construct a region of interest coverage plot</i> |
|--------|---|

Description

Given a list of data frames construct a sequencing coverage view over a region of interest.

Usage

```
genCov(x, txdb, gr, genome, reduce = FALSE, gene_colour = NULL,
  gene_name = "Gene", gene_plotLayer = NULL, label_bgFill = "black",
  label_txtFill = "white", label_borderFill = "black",
  label_txtSize = 10, lab2plot_ratio = c(1, 10), cov_colour = "blue",
  cov_plotType = "point", cov_plotLayer = NULL, base = c(10, 2, 2),
  transform = c("Intron", "CDS", "UTR"), gene_labelTranscript = TRUE,
  gene_labelTranscriptSize = 4, gene_isoformSel = NULL, out = "plot",
  subsample = FALSE)
```

Arguments

| | |
|---|---|
| x | Named list with list elements containing data frames representing samples. Data frame rows should represent read pileups observed in sequencing data. Data frame column names must include "end" and "cov" corresponding to the base end position and coverage of a pileup respectively. Data within data frames must be on the same chromosome as the region of interest, see details! |
|---|---|

| | |
|--------------------------|--|
| txdb | Object of class TxDb giving transcription meta data for a genome assembly. See Bioconductor annotation packages. |
| gr | Object of class GRanges specifying the region of interest and corresponding to a single gene. See Bioconductor package GRanges. |
| genome | Object of class BSgenome specifying the genome sequence of interest. See Bioconductor annotation packages. |
| reduce | Boolean specifying whether to collapse gene isoforms within the region of interest into one representative transcript. Experimental use with caution! |
| gene_colour | Character string specifying the colour of the gene to be plotted in the gene track. |
| gene_name | Character string specifying the name of the gene or region of interest. |
| gene_plotLayer | Valid ggplot2 layer to be added to the gene sub-plot. |
| label_bgFill | Character string specifying the desired background colour of the track labels. |
| label_txtFill | Character string specifying the desired text colour of the track labels. |
| label_borderFill | Character string specifying the desired border colour of the track labels. |
| label_txtSize | Integer specifying the size of the text within the track labels. |
| lab2plot_ratio | Numeric vector of length 2 specifying the ratio of track labels to plot space. |
| cov_colour | Character string specifying the colour of the data in the coverage plots. |
| cov_plotType | Character string specifying one of "line", "bar" or "point". Changes the ggplot2 geom which constructs the data display. |
| cov_plotLayer | Valid ggplot2 layer to be added to the coverage sub-plots. |
| base | Numeric vector of log bases to transform the data corresponding to the elements supplied to the variable transform See details. |
| transform | Character vector specifying what objects to log transform, accepts "Intron", "CDS", and "UTR" See details. |
| gene_labelTranscript | Boolean specifying whether to plot the transcript names in the gene plot. |
| gene_labelTranscriptSize | Integer specifying the size of the transcript name text in the gene plot. |
| gene_isoformSel | Character vector specifying the names (from the txdb object) of isoforms within the region of interest to display. |
| out | Character vector specifying the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |
| subsample | Boolean value specifying whether to reduce the provided coverage data to a subset of approximately 1000 points. Used to generate sparse plots that use less disk space and are faster to render. |

Details

genCov is a function designed construct a series of tracks based on a TxDb object giving transcript features, and coverage data supplied to parameter 'x'. The function will look at a region of interest specified by the argument supplied to gr and plot transcript features and the corresponding coverage information. The argument supplied to 'genome' enables gc content within genomic features to be calculated and displayed. The argument supplied to x must contain data on the same chromosome as the region of interest specified in the parameter 'gr'!

Typically, introns of a transcript are much larger than exons, while exons are sometimes of greater interest. To address this, `genCov` will by default scale the x-axis to expand track information according to region type: coding sequence (CDS), untranslated region (UTR), or intron / intergenic (Intron). The amount by which each region is scaled is controlled by the `'base'` and `'transform'` arguments. `'transform'` specifies which regions to scale, and `'base'` corresponds to the log base transform to apply to those regions. To keep one or more region types from being scaled, omit the corresponding entries from the `'base'` and `'transform'` vectors.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Load transcript meta data
library(TxDb.Hsapiens.UCSC.hg19.knownGene)
txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene

# Load BSgenome
library(BSgenome.Hsapiens.UCSC.hg19)
genome <- BSgenome.Hsapiens.UCSC.hg19

# Define a region of interest
gr <- GRanges(seqnames=c("chr10"),
              ranges=IRanges(start=c(89622195), end=c(89729532)), strand=strand(c("+")))

# Create Data for input
start <- c(89622194:89729524)
end <- c(89622195:89729525)
chr <- 10
cov <- c(rnorm(100000, mean=40), rnorm(7331, mean=10))
cov_input_A <- as.data.frame(cbind(chr, start, end, cov))

start <- c(89622194:89729524)
end <- c(89622195:89729525)
chr <- 10
cov <- c(rnorm(50000, mean=40), rnorm(7331, mean=10), rnorm(50000, mean=40))
cov_input_A <- as.data.frame(cbind(chr, start, end, cov))

# Define the data as a list
data <- list("Sample A"=cov_input_A)

# Call genCov
genCov(data, txdb, gr, genome, gene_labelTranscriptSize=3)
```

geneViz

Construct a gene-features plot

Description

Given a `GRanges` object specifying a region of interest, plot genomic features within that region.

Usage

```
geneViz(txdb, gr, genome, reduce = FALSE, gene_colour = NULL,
        base = c(10, 2, 2), transform = c("Intron", "CDS", "UTR"),
        isoformSel = NULL, labelTranscript = TRUE, labelTranscriptSize = 4,
        plotLayer = NULL)
```

Arguments

| | |
|---------------------|---|
| txdb | Object of class TxDb giving transcription meta data for a genome assembly. See Bioconductor annotation packages. |
| gr | Object of class GRanges specifying the region of interest and corresponding to a single gene. See Bioconductor package GRanges. |
| genome | Object of class BSgenome specifying the genome sequence of interest. See Bioconductor annotation packages. |
| reduce | Boolean specifying whether to collapse gene isoforms within the region of interest into one representative transcript. Experimental use with caution! |
| gene_colour | Character string specifying the colour of the gene to be plotted. |
| base | Numeric vector of log bases to transform the data corresponding to the elements supplied to the variable transform See details. |
| transform | Character vector specifying what objects to log transform, accepts "Intron", "CDS", and "UTR" See details. |
| isoformSel | Character vector specifying the names (from the txdb object) of isoforms within the region of interest to display. |
| labelTranscript | Boolean specifying whether to plot the transcript names in the gene plot. |
| labelTranscriptSize | Integer specifying the size of the transcript name text in the gene plot. |
| plotLayer | Valid ggplot2 layer to be added to the gene plot. |

Details

geneViz is an internal function which will output a list of three elements. As a convenience the function is exported however to obtain the plot from geneViz the user must call the first element of the list. geneViz is intended to plot gene features within a single gene with boundaries specified by the GRanges object, plotting more than one gene is advised against.

Typically, introns of a transcript are much larger than exons, while exons are sometimes of greater interest. To address this, geneCov will by default scale the x-axis to expand track information according to region type: coding sequence (CDS), untranslated region (UTR), or intron / intergenic (Intron). The amount by which each region is scaled is controlled by the 'base' and 'transform' arguments. 'transform' specifies which regions to scale, and 'base' corresponds to the log base transform to apply to those regions. To keep one or more region types from being scaled, omit the corresponding entries from the 'base' and 'transform' vectors.

Value

object of class list with list elements containing a ggplot object, the gene features within the plot as a data frame, and mapping information of the gene features within the ggplot object.

Examples

```
# need transcript data for reference
library(TxDb.Hsapiens.UCSC.hg19.knownGene)
txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene

# need a biostrings object for reference
library(BSgenome.Hsapiens.UCSC.hg19)
genome <- BSgenome.Hsapiens.UCSC.hg19

# need Granges object
gr <- GRanges(seqnames=c("chr10"),
              ranges=IRanges(start=c(89622195), end=c(89729532)), strand=strand(c("+")))

# Plot the graphic
geneViz(txdb, gr, genome)
```

GenVisR

*GenVisR***Description**

GenVisR

getData

*Method getData***Description**

Method getData

Helper function to get data from classes

Helper function to getData from classes, under development!!!

Helper function to get data from classes

Helper function to getData from classes, under development!!!

Usage

getData(object, ...)

S4 method for signature 'Clinical'

getData(object, ...)

S4 method for signature 'ClinicalData'

getData(object, ...)

.getData_Lollipop(object, name = NULL, index = NULL, ...)

S4 method for signature 'LollipopData'

getData(object, name = NULL, index = NULL,
...)

```

## S4 method for signature 'Lollipop'
getData(object, name = NULL, index = NULL, ...)

.getData_MutSpectra(object, name = NULL, index = NULL, ...)

## S4 method for signature 'MutSpectraPrimaryData'
getData(object, name = NULL,
        index = NULL, ...)

## S4 method for signature 'MutSpectra'
getData(object, name = NULL, index = NULL, ...)

.getData_Rainfall(object, name = NULL, index = NULL, ...)

## S4 method for signature 'RainfallPrimaryData'
getData(object, name = NULL,
        index = NULL, ...)

## S4 method for signature 'Rainfall'
getData(object, name = NULL, index = NULL, ...)

.getData_waterfall(object, name = NULL, index = NULL, ...)

## S4 method for signature 'WaterfallData'
getData(object, name = NULL, index = NULL,
        ...)

## S4 method for signature 'Waterfall'
getData(object, name = NULL, index = NULL, ...)

```

Arguments

| | |
|--------|--|
| object | Object of class Clinical, |
| ... | additional arguments to passed |
| name | String corresponding to the slot for which to extract data from. |
| index | Integer specifying the slot for which to extract data from. |

Details

The `getData` method is an accessor function used to access data held in GenVisR objects.

| | |
|----------------|------------------------------|
| getDescription | <i>Method getDescription</i> |
|----------------|------------------------------|

Description

Method `getDescription`

Usage

```
getDescription(object, ...)

## S4 method for signature 'VEP_Virtual'
getDescription(object, ...)

## S4 method for signature 'VEP'
getDescription(object, ...)
```

Arguments

| | |
|--------|--------------------------------|
| object | Object of class VEP |
| ... | additional arguments to passed |

| | |
|---------|-----------------------|
| getGrob | <i>Method getGrob</i> |
|---------|-----------------------|

Description

Method getGrob

Usage

```
getGrob(object, ...)

## S4 method for signature 'LolliplotPlots'
getGrob(object, index = 1, ...)

## S4 method for signature 'Lolliplot'
getGrob(object, index = 1, ...)

## S4 method for signature 'MutSpectraPlots'
getGrob(object, index = 1, ...)

## S4 method for signature 'MutSpectra'
getGrob(object, index = 1, ...)

## S4 method for signature 'RainfallPlots'
getGrob(object, index = 1, ...)

## S4 method for signature 'Rainfall'
getGrob(object, index = 1, ...)

## S4 method for signature 'WaterfallPlots'
getGrob(object, index = 1, ...)

## S4 method for signature 'Waterfall'
getGrob(object, index = 1, ...)
```

Arguments

| | |
|--------|--|
| object | Object of clas MutSpectra |
| ... | additional arguments to passed |
| index | integer specifying the plot index to extract |

| | |
|-----------|-------------------------|
| getHeader | <i>Method getHeader</i> |
|-----------|-------------------------|

Description

Method getHeader

Usage

```

getHeader(object, ...)

## S4 method for signature 'VEP_Virtual'
getHeader(object, ...)

## S4 method for signature 'VEP'
getHeader(object, ...)

```

Arguments

| | |
|--------|--------------------------------|
| object | Object of class VEP |
| ... | additional arguments to passed |

| | |
|---------|-----------------------|
| getMeta | <i>Method getMeta</i> |
|---------|-----------------------|

Description

Method getMeta

Usage

```

getMeta(object, ...)

## S4 method for signature 'GMS_Virtual'
getMeta(object, ...)

## S4 method for signature 'GMS'
getMeta(object, ...)

## S4 method for signature 'MutationAnnotationFormat_Virtual'
getMeta(object, ...)

## S4 method for signature 'MutationAnnotationFormat'

```

```

getMeta(object, ...)

## S4 method for signature 'VEP_Virtual'
getMeta(object, ...)

## S4 method for signature 'VEP'
getMeta(object, ...)

```

Arguments

| | |
|--------|---|
| object | Object of class VEP, GMS, or MutationAnnotationFormat |
| ... | additional arguments to passed |

| | |
|-------------|---------------------------|
| getMutation | <i>Method getMutation</i> |
|-------------|---------------------------|

Description

Method getMutation

Usage

```

getMutation(object, ...)

## S4 method for signature 'GMS_Virtual'
getMutation(object, ...)

## S4 method for signature 'GMS'
getMutation(object, ...)

## S4 method for signature 'MutationAnnotationFormat_Virtual'
getMutation(object, ...)

## S4 method for signature 'MutationAnnotationFormat'
getMutation(object, ...)

## S4 method for signature 'VEP_Virtual'
getMutation(object, ...)

## S4 method for signature 'VEP'
getMutation(object, ...)

```

Arguments

| | |
|--------|---|
| object | Object of class VEP, GMS, or MutationAnnotationFormat |
| ... | additional arguments to passed |

| | |
|---------|-----------------------|
| getPath | <i>Method getPath</i> |
|---------|-----------------------|

Description

Method getPath

Usage

```
getPath(object, ...)
```

```
## S4 method for signature 'GMS'  
getPath(object, ...)
```

```
## S4 method for signature 'MutationAnnotationFormat'  
getPath(object, ...)
```

```
## S4 method for signature 'VEP'  
getPath(object, ...)
```

Arguments

| | |
|--------|---|
| object | Object of class VEP, GMS, or MutationAnnotationFormat |
| ... | additional arguments to passed |

| | |
|-------------|---------------------------|
| getPosition | <i>Method getPosition</i> |
|-------------|---------------------------|

Description

Method getPosition

Usage

```
getPosition(object, ...)
```

```
## S4 method for signature 'GMS_Virtual'  
getPosition(object, ...)
```

```
## S4 method for signature 'GMS'  
getPosition(object, ...)
```

```
## S4 method for signature 'MutationAnnotationFormat_Virtual'  
getPosition(object, ...)
```

```
## S4 method for signature 'MutationAnnotationFormat'  
getPosition(object, ...)
```

```
## S4 method for signature 'VEP_Virtual'
```

```
getPosition(object, ...)

## S4 method for signature 'VEP'
getPosition(object, ...)
```

Arguments

| | |
|--------|---|
| object | Object of class VEP, GMS, or MutationAnnotationFormat |
| ... | additional arguments to passed |

| | |
|-----------|-------------------------|
| getSample | <i>Method getSample</i> |
|-----------|-------------------------|

Description

Method getSample

Usage

```
getSample(object, ...)

## S4 method for signature 'GMS_Virtual'
getSample(object, ...)

## S4 method for signature 'GMS'
getSample(object, ...)

## S4 method for signature 'MutationAnnotationFormat_Virtual'
getSample(object, ...)

## S4 method for signature 'MutationAnnotationFormat'
getSample(object, ...)

## S4 method for signature 'VEP_Virtual'
getSample(object, ...)

## S4 method for signature 'VEP'
getSample(object, ...)
```

Arguments

| | |
|--------|---|
| object | Object of class VEP, GMS, or MutationAnnotationFormat |
| ... | additional arguments to passed |

| | |
|------------|--------------------------|
| getVersion | <i>Method getVersion</i> |
|------------|--------------------------|

Description

Method getVersion

Usage

```
getVersion(object, ...)
```

```
## S4 method for signature 'GMS'
getVersion(object, ...)
```

```
## S4 method for signature 'MutationAnnotationFormat'
getVersion(object, ...)
```

```
## S4 method for signature 'VEP'
getVersion(object, ...)
```

Arguments

| | |
|--------|---|
| object | Object of class VEP, GMS, or MutationAnnotationFormat |
| ... | additional arguments to passed |

| | |
|-----------|------------------|
| GMS-class | <i>Class GMS</i> |
|-----------|------------------|

Description

An S4 class for Genome Modeling System annotation files, under development!!!

Usage

```
GMS(path, data = NULL, version = 4, verbose = FALSE)
```

Arguments

| | |
|---------|---|
| path | String specifying the path to a GMS annotation file. Can accept wildcards if multiple GMS annotation files exist (see details). |
| data | data.table object storing a GMS annotation file. Overrides "path" if specified. |
| version | String specifying the version of the GMS files, Defaults to version 4. |
| verbose | Boolean specifying if progress should be reported while reading in the GMS files. |

Details

When specifying a path to a GMS annotation file the option exist to either specify the full path to an annotation file or to use wildcards to specify multiple files. When specifying a full path the initializer will check if a column named "sample" containing the relevant sample for each row exists. If such a column is not found the initializer will assume this file corresponds to only one sample and populate a sample column accordingly. Alternatively if multiple files are specified at once using a wildcard, the initializer will aggregate all the files and use the file names minus any extension to populate sample names. The version defaults to 4 which is the default value of the GMS annotator. This value will need to be changed only if files were created using a different GMS annotator version.

Slots

`path` Character string specifying the paths of the GMS files read in.

`version` Numeric value specifying the version of the GMS annotation files.

`gmsObject` gms object which inherits from `gms_Virtual` class.

See Also

[Waterfall](#)

[MutSpectra](#)

GMS_v4-class

Class GMS_v4

Description

An S4 class to represent data in gms annotation version 4, inherits from the `GMS_Virtual` class.

Usage

```
GMS_v4(gmsData)
```

Arguments

`gmsData` data.table object containing a gms annotation file conforming to the version 4 specifications.

Slots

`position` data.table object containing column names "chromosome_name", "start", "stop".

`mutation` data.table object containing column names "reference", "variant", "trv_type".

`sample` data.table object containing columns names "sample".

`meta` data.table object containing meta data.

| | |
|-------------------|--------------------------|
| GMS_Virtual-class | <i>Class GMS_Virtual</i> |
|-------------------|--------------------------|

Description

An S4 class to act as a virtual class for GMS version sub-classes.

Slots

position data.table object holding genomic positions.
 mutation data.table object holding mutation status data.
 sample data.table object holding sample data.
 meta data.table object holding all other meta data.

| | |
|------------------|-----------------------|
| HCC1395_Germline | <i>Germline Calls</i> |
|------------------|-----------------------|

Description

A data set containing downsampled Germline calls originating from the HCC1395 breast cancer cell line.

Usage

```
data(HCC1395_Germline)
```

Format

a data frame with 9200 observations and 5 variables

Value

Object of class data frame

| | |
|-----------|-------------------|
| HCC1395_N | <i>Normal BAM</i> |
|-----------|-------------------|

Description

A data set containing read pileups intersecting 24 identity snp locations from GenVisR::SNPloci. Pileups are from downsampled bams and originate from normal tissue corresponding to the HCC1395 breast cancer cell line.

Usage

```
data(HCC1395_N)
```

Format

a data frame with 59 observations and 6 variables

Value

Object of class list

HCC1395_T

Tumor BAM

Description

A data set containing read pileups intersecting 24 identity snp locations from GenVisR::SNPloci. Pileups are from downsampled bams and originate from tumor tissue corresponding to the HCC1395 breast cancer cell line.

Usage

data(HCC1395_T)

Format

a data frame with 52 observations and 6 variables

Value

Object of class list

hg19chr

hg19 chromosome boundaries

Description

A data set containing chromosome boundaries corresponding to hg19.

Usage

data(hg19chr)

Format

a data frame with 24 observations and 3 variables

Value

Object of class data frame

| | |
|----------|------------------------------|
| ideoView | <i>Construct an ideogram</i> |
|----------|------------------------------|

Description

Given a data frame with cytogenetic information, construct an ideogram.

Usage

```
ideoView(x, chromosome = "chr1", txtAngle = 45, txtSize = 5,  
         plotLayer = NULL, out = "plot")
```

Arguments

| | |
|------------|---|
| x | Object of class data frame with rows representing cytogenetic bands. The data frame must contain the following column names "chrom", "chromStart", "chromEnd", "name", "gieStain" |
| chromosome | Character string specifying which chromosome from the "chrom" column in the argument supplied to parameter x to plot. |
| txtAngle | Integer specifying the angle of text labeling cytogenetic bands. |
| txtSize | Integer specifying the size of text labeling cytogenetic bands. |
| plotLayer | additional ggplot2 layers for the ideogram |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |

Details

ideoView is a function designed to plot cytogenetic band information. Modifications to the graphic object can be made via the 'plotLayer' parameter, see vignette for details.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Obtain cytogenetic information for the genome of interest from attached  
# data set cytoGeno  
data <- cytoGeno[cytoGeno$genome == 'hg38',]  
  
# Call ideoView for chromosome 1  
ideoView(data, chromosome='chr1', txtSize=4)
```

lohSpec

*Plot LOH data***Description**

Construct a graphic visualizing Loss of Heterozygosity in a cohort

Usage

```
lohSpec(x = NULL, path = NULL, fileExt = NULL, y = NULL,
        genome = "hg19", gender = NULL, step = 1e+06,
        window_size = 2500000, normal = 0.5, colourScheme = "inferno",
        plotLayer = NULL, method = "slide", out = "plot")
```

Arguments

| | |
|--------------|---|
| x | object of class data frame with rows representing germline calls. The data frame must contain columns with the following names "chromosome", "position", "n_vaf", "t_vaf", "sample". required if path is set to NULL (see details). vaf should range from 0-1. |
| path | Character string specifying the path to a directory containing germline calls for each sample. Germline calls are expected to be stored as tab-separated files which contain the following column names "chromosome", "position", "n_vaf", "t_vaf", and "sample". required if x is set to null (see details). |
| fileExt | Character string specifying the file extensions of files within the path specified. Required if argument is supplied to path (see details). |
| y | Object of class data frame with rows representing chromosome boundaries for a genome assembly. The data frame must contain columns with the following names "chromosome", "start", "end" (optional: see details). |
| genome | Character string specifying a valid UCSC genome (see details). |
| gender | Character vector of length equal to the number of samples, consisting of elements from the set "M", "F". Used to suppress the plotting of allosomes where appropriate. |
| step | Integer value specifying the step size (i.e. the number of base pairs to move the window). required when method is set to slide (see details). |
| window_size | Integer value specifying the size of the window in base pairs in which to calculate the mean Loss of Heterozygosity (see details). |
| normal | Numeric value within the range 0-1 specifying the expected normal variant allele frequency to be used in Loss of Heterozygosity calculations. defaults to .50% |
| colourScheme | Character vector specifying the colour scale to use from the viridis package. One of "viridis", "magma", "plasma", or "inferno". |
| plotLayer | Valid ggplot2 layer to be added to the plot. |
| method | character string specifying the approach to be used for displaying Loss of Heterozygosity, one of "tile" or "slide" (see details). |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |

Details

lohSpec is intended to plot the loss of heterozygosity (LOH) within a sample. As such lohSpec expects input data to contain only LOH calls. Input can be supplied as a single data frame given to the argument `x` with rows containing germline calls and variables giving the chromosome, position, normal variant allele frequency, tumor variant allele frequency, and the sample. In lieu of this format a series of .tsv files can be supplied via the `path` and `fileExt` arguments. If this method is chosen samples will be inferred from the file names. In both cases columns containing the variant allele frequency for normal and tumor samples should range from 0-1. Two methods exist to calculate and display LOH events. If the method is set to "tile" mean LOH is calculated based on the `window_size` argument with windows being placed next to each other. If the method is set to slide the window will slide and calculate the LOH based on the `step` parameter. In order to ensure the entire chromosome is plotted lohSpec requires the location of chromosome boundaries for a given genome assembly. As a convenience this information is available for the following genomes "hg19", "hg38", "mm9", "mm10", "rn5" and can be retrieved by supplying one of the aforementioned assemblies via the 'genome' parameter. If an argument is supplied to the 'genome' parameter and is unrecognized a query to the UCSC MySQL database will be attempted to obtain the required information. If chromosome boundary locations are unavailable for a given assembly this information can be supplied to the 'y' parameter which has priority over the 'genome' parameter.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# plot loh within the example dataset
lohSpec(x=HCC1395_Germline)
```

 lohView

Construct LOH chromosome plot

Description

Given a data frame construct a plot to display Loss of Heterozygosity for specific chromosomes.

Usage

```
lohView(x, y = NULL, genome = "hg19", chr = "chr1",
        ideogram_txtAngle = 45, ideogram_txtSize = 5, plotLayer = NULL,
        ideogramLayer = NULL, out = "plot")
```

Arguments

- | | |
|---------------------|---|
| <code>x</code> | object of class data frame with rows representing Heterozygous Germline calls. The data frame must contain columns with the following names "chromosome", "position", "n_vaf", "t_vaf", "sample". |
| <code>y</code> | Object of class data frame with rows representing cytogenetic bands for a chromosome. The data frame must contain columns with the following names "chrom", "chromStart", "chromEnd", "name", "gieStain" for plotting the ideogram (optional: see details). |
| <code>genome</code> | Character string specifying a valid UCSC genome (see details). |

| | |
|-------------------|---|
| chr | Character string specifying which chromosome to plot one of "chr..." or "all" |
| ideogram_txtAngle | Integer specifying the angle of cytogenetic labels on the ideogram subplot. |
| ideogram_txtSize | Integer specifying the size of cytogenetic labels on the ideogram subplot. |
| plotLayer | Valid ggplot2 layer to be added to the copy number plot. |
| ideogramLayer | Valid ggplot2 layer to be added to the ideogram sub-plot. |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |

Details

lohView is able to plot in two modes specified via the 'chr' parameter, these modes are single chromosome view in which an ideogram is displayed and genome view where chromosomes are faceted. For the single chromosome view cytogenetic band information is required giving the coordinate, stain, and name of each band. As a convenience GenVisR stores this information for the following genomes "hg19", "hg38", "mm9", "mm10", and "rn5". If the genome assembly supplied to the 'genome' parameter is not one of the 5 afore mentioned genome assemblies GenVisR will attempt to query the UCSC MySQL database to retrieve this information. Alternatively the user can manually supply this information as a data frame to the 'y' parameter, input to the 'y' parameter take precedence of input to 'genome'.

A word of caution, users are advised to only use heterozygous germline calls in input to 'x', failure to do so may result in a misleading visual!

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Plot loh for chromosome 5
lohView(HCC1395_Germline, chr='chr5', genome='hg19', ideogram_txtSize=4)
```

| | |
|----------|-----------------------------|
| lollipop | <i>Construct a lollipop</i> |
|----------|-----------------------------|

Description

Given a data frame construct a plot displaying mutations on a transcript framework.

Usage

```
lollipop(x, y = NULL, z = NULL, fillCol = NULL, labelCol = NULL,
  txtAngle = 45, txtSize = 5, pntSize = 4,
  proteinColour = "#999999", obsA.rep.fact = 5000,
  obsA.rep.dist.lmt = 500, obsA.attr.fact = 0.1, obsA.adj.max = 0.1,
  obsA.adj.lmt = 0.5, obsA.iter.max = 50000, obsB.rep.fact = 5000,
  obsB.rep.dist.lmt = 500, obsB.attr.fact = 0.1, obsB.adj.max = 0.1,
  obsB.adj.lmt = 0.5, obsB.iter.max = 50000, sideChain = FALSE,
  species = "hsapiens", maxLolliStack = NULL, plotLayer = NULL,
  paletteA = NULL, paletteB = NULL, host = "www.ensembl.org",
  out = "plot")
```

Arguments

| | |
|-------------------|--|
| x | Object of class data frame with rows representing mutations. The data frame must contain columns with the following names "transcript_name", "gene", and "amino_acid_change". Values in the "transcript_name" column must represent an ensembl transcript id and values in the "amino_acid_change" column must be in p.notation (see details). |
| y | Object of class data frame with rows representing mutations. The data frame must contain columns with the following names "transcript_name" and "amino_acid_change". Values in the "transcript_name" column must represent an ensembl transcript id and values in the "amino_acid_change" column must be in p. notation (optional, see details). |
| z | Object of class data frame with rows representing regions of interest. The data frame must contain columns with the following names "description", "start", "stop" (optional see details). |
| fillCol | Character string specifying the column name of the argument supplied to parameter x on which to colour the lollis representing mutations (see details). |
| labelCol | Character string specifying the column name of the argument supplied to parameter x from which to extract and display text corresponding to mutations (see details). |
| txtAngle | Integer specifying the angle of label text to be plotted if an argument is supplied to the labelCol parameter. |
| txtSize | Integer specifying the size of label text to be plotted if an argument is supplied to the labelCol parameter. |
| pntSize | Integer specifying the size of lolli points representing mutations. |
| proteinColour | Character string specifying the background colour of the protein. |
| obsA.rep.fact | Numeric value representing the repulsive factor for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette). |
| obsA.rep.dist.lmt | Numeric value representing the repulsive distance limit for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette). |
| obsA.attr.fact | Numeric value representing the attraction factor for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette). |
| obsA.adj.max | Numeric value representing the max position adjustment for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette). |
| obsA.adj.lmt | Numeric value representing the adjustment limit for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette). |
| obsA.iter.max | Integer representing the number of iterations of position adjustments for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette). |
| obsB.rep.fact | Numeric value representing the repulsive factor for the lollis plotted, which were derived from the argument supplied to parameter y (see details and vignette). |
| obsB.rep.dist.lmt | Numeric value representing the repulsive distance limit for the lollis plotted, which were derived from the argument supplied to parameter y (see details and vignette). |

| | |
|-----------------------------|---|
| <code>obsB.attr.fact</code> | Numeric value representing the attraction factor for the lollis plotted, which were derived from the argument supplied to parameter <code>y</code> (see details and vignette). |
| <code>obsB.adj.max</code> | Numeric value representing the max position adjustment for the lollis plotted, which were derived from the argument supplied to parameter <code>y</code> (see details and vignette). |
| <code>obsB.adj.lmt</code> | Numeric value representing the adjustment limit for the lollis plotted, which were derived from the argument supplied to parameter <code>y</code> (see details and vignette). |
| <code>obsB.iter.max</code> | Integer representing the number of iterations of position adjustments for the lollis plotted, which were derived from the argument supplied to parameter <code>y</code> (see details and vignette). |
| <code>sideChain</code> | Boolean specifying if amino acid sidechain data should be plotted in lieu of protein domains (see details). |
| <code>species</code> | A valid species from which to retrieve protein domain and sequence data for a given transcript (see details). |
| <code>maxLolliStack</code> | Integer specifying the cutoff for the maximum number of lollis allowed to be stacked at a single position. |
| <code>plotLayer</code> | Valid ggplot2 layer to be added to the plot. |
| <code>paletteA</code> | Character vector specifying colours for protein domains, valid only if <code>sideChain==FALSE</code> . |
| <code>paletteB</code> | Character vector specifying colours for lollis representing mutations, valid only if argument is supplied to <code>fillCol</code> . |
| <code>host</code> | Host to connect to for biomaRt queries (see details). |
| <code>out</code> | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |

Details

lollipop is a function designed to display mutation information in the context of a protein identified by an ensembl transcript id. The lollipop function will query ensembl via biomart to retrieve sequence and domain information in order to construct a representation of a protein and therefore requires an internet connection. A value must be supplied to the `species` parameter (defaults to `hsapiens`) in order for a successful biomart query. Valid arguments to this field are those species with datasets available via ensembl. please specify species in lowercase without a period (i.e. `hsapiens` instead of `H.sapiens`), lollipop will inform the user of available species if input to the `species` parameter is not recognized. Further lollipop will build a protein framework based on sequence data obtained from biomaRt, by default this will default to the latest ensembl version. In order for the most accurate representation the annotation version of the mutations given to lollipop should match the annotation version used by biomaRt. The annotation version used by biomaRt can be changed via the `host` parameter (see vignette for more details).

lollipop is capable of plotting two separate sets of data on the protein representation specified by parameters `'x'` and `'y'`, the data supplied to these parameters will be plotted on the top and bottom of the protein respectively. Note that input to these parameters is expected to correspond to a single ensembl transcript and that values in the "amino_acid_change" columns are required to be in p. notation (i.e. `p.V600E`). Further lollipop is able to plot custom domain annotation if supplied via the parameter `'z'`, this will override domain information obtained from biomart.

lollipop uses a forcefield model from the package `FField` to attract and repulse lollis. The parameters for this force field model are set to reasonable defaults however may be adjusted via the `obsA...` and `obsB...` family of parameters. Please see the package `FField` available on cran for a description

of these parameters. Note that the time to construct the lollipop will in large part depend on the number of mutations and the values supplied to the forcefield parameters.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Create input data
data <- brcaMAF[brcaMAF$Hugo_Symbol == 'TP53',c('Hugo_Symbol', 'amino_acid_change_WU')]
data <- as.data.frame(cbind(data, 'ENST00000269305'))
colnames(data) <- c('gene', 'amino_acid_change', 'transcript_name')

# Call lollipop
lollipop(data)
```

| | |
|----------------|-----------------------|
| Lollipop-class | <i>Class Lollipop</i> |
|----------------|-----------------------|

Description

An S4 class for the lollipop object, under development!!!

Usage

```
Lollipop(input, transcript = NULL, species = "hsapiens",
  host = "www.ensembl.org", txdb = NULL, BSgenome = NULL,
  emphasize = NULL, DomainPalette = NULL, MutationPalette = NULL,
  labelAA = TRUE, plotALayers = NULL, plotBLayers = NULL,
  sectionHeights = NULL, verbose = FALSE)
```

Arguments

| | |
|-----------------|---|
| input | Object of class MutationAnnotationFormat, GMS, VEP, or a data.table with appropriate columns |
| transcript | Character string specifying the ensembl transcript for which to plot, should be a transcript which corresponds to the gene parameter. |
| species | Character string specifying a species when using biomaRt queries |
| host | Character string specifying a host to connect to when using biomaRt queries |
| txdb | A bioconductor txdb object to annotate amino acid positions, required only if amino acid changes are missing (see details). |
| BSgenome | A bioconductor BSgenome object to annotate amino acid positions, required only if amino acid changes are missing (see details). |
| emphasize | Character vector specifying a list of mutations to emphasize. |
| DomainPalette | Character vector specifying the colors used for encoding protein domains |
| MutationPalette | Character vector specifying the colors used for encoding mutations |
| labelAA | Boolean specifying if labels should be added to emphasized mutations |

plotALayers list of ggplot2 layers to be passed to the density plot.
 plotBLayers list of ggplot2 layers to be passed to the lollipop.
 sectionHeights Numeric vector specifying relative heights of each plot section, should sum to one. Expects a value for each section.
 verbose Boolean specifying if status messages should be reported.

Slots

PlotA gtable object for the top sub-plot
 PlotB gtable object for the bottom sub-plot
 Grob gtable object storing the arranged plot
 primaryData data.table object storing the primary data
 geneData data.table object storing gene and domain coordinates

Examples

```

# Load a pre-existing data set
dataset <- PIK3CA

# mode 1, amino acid changes are not present

library(TxDb.Hsapiens.UCSC.hg38.knownGene)
library(BSgenome.Hsapiens.UCSC.hg38)
txdb <- TxDb.Hsapiens.UCSC.hg38.knownGene
BSgenome <- BSgenome.Hsapiens.UCSC.hg38

keep <- c("Chromosome", "Start_Position", "End_Position", "Reference_Allele",
          "Tumor_Seq_Allele2", "Tumor_Sample_Barcode", "Gene", "Variant_Classification")
dataset.mode1 <- dataset[,keep]
colnames(dataset.mode1) <- c("chromosome", "start", "stop", "reference", "variant",
                             "sample", "gene", "consequence")

# mode 2, amino acid changes are present

keep <- c("Chromosome", "Start_Position", "End_Position", "Reference_Allele",
          "Tumor_Seq_Allele2", "Tumor_Sample_Barcode", "Gene", "Variant_Classification",
          "Transcript_ID", "HGVS")
dataset.mode2 <- dataset[,keep]
colnames(dataset.mode2) <- c("chromosome", "start", "stop", "reference", "variant",
                             "sample", "gene", "consequence", "transcript", "proteinCoord")

# run Lollipop

object <- Lollipop(dataset.mode1, transcript="ENST00000263967",
                  species="hsapiens", txdb=txdb, BSgenome=BSgenome)
object <- Lollipop(dataset.mode2, transcript="ENST00000263967",
                  species="hsapiens")

```

 lollipop_AA2sidechain

Convert AA to side chain classification

Description

Given the 1 letter code an amino acid, return the side chain classification

Usage

```
lollipop_AA2sidechain(x)
```

Arguments

x Character of length 1 giving the 1 letter amino acid code

Value

Object of class character

lollipop_buildMain

Construct Lollipop

Description

Construct Lollipop given gene and mutation data

Usage

```
lollipop_buildMain(gene_data, length, mutation_observed,
  mutation_observed2, fill_value, label_column, plot_text_angle,
  plot_text_size, point_size, gene_colour, sequence_data,
  plot_sidechain = FALSE, layers = NULL, paletteA = NULL,
  paletteB = NULL)
```

Arguments

gene_data object of class dataframe giving protien domain and gene information

length integer specifying the length of the protien in amino acids

mutation_observed object of class data frame specifying mutations observed in input file

mutation_observed2 optional object of class data frame specifying additional mutations for bottom track

fill_value character string specifying the column on which to colour mutation points

label_column character string specifying the column containing the labels to attach to mutation points

| | |
|-----------------|---|
| plot_text_angle | numeric value specifying the angle of text to be plotted |
| plot_text_size | numeric value specifying the size of text to be plotted |
| point_size | numeric value specifying the size of mutation points |
| gene_colour | color to shade plotted gene |
| sequence_data | object of class dataframe giving AA sequence, sidechain, and coord required if plot_sidechain is true |
| plot_sidechain | boolean specifying whether to plot the AA sidechain instead of domain information |
| layers | additional ggplot2 layers to plot |
| paletteA | Character vector specifying colours for gene features |
| paletteB | Character vector specifying colours for lolli features |

Value

a ggplot2 object

LucCNseg

Truncated CN segments

Description

A data set in long format containing Copy Number segments for 4 samples corresponding to "lung cancer" from Govindan et al. Cell. 2012, PMID:22980976

Usage

```
data(LucCNseg)
```

Format

a data frame with 3336 observations and 6 variables

Value

Object of class data frame

 MutationAnnotationFormat-class

Class MutationAnnotationFormat

Description

An S4 class acting as a container for MutationAnnotationFormat version sub-classes, under development!!!

Usage

```
MutationAnnotationFormat(path, version = "auto", verbose = FALSE)
```

Arguments

| | |
|---------|---|
| path | String specifying the path to a MAF file. |
| version | String specifying the version of the MAF file, if set to auto the version will be obtained from the header in the MAF file. |
| verbose | Boolean specifying if progress should be reported while reading in the MAF file. |

Slots

| | |
|-----------|---|
| path | Character string specifying the path of the MAF file read in. |
| version | Numeric value specifying the version of the MAF file. |
| mafObject | MutationAnnotationFormat object which inherits from MutationAnnotationFormat_Virtual class. |

See Also

[Waterfall](#)
[MutSpectra](#)

 MutationAnnotationFormat_v1.0-class

Class MutationAnnotationFormat_v1.0

Description

An S4 class to represent data in mutation annotation format version 1.0, inherits from the MutationAnnotationFormat_Virtual class.

Usage

```
MutationAnnotationFormat_v1.0(mafData)
```

Arguments

mafData data.table object containing a maf file conforming to the version 1.0 specification.

Slots

position data.table object containing column names "Chromosome", "Start_Position", "End_Position", "Strand".

mutation data.table object containing column names "Variant_Classification", "Variant_Type", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2".

sample data.table object containing columns names "Tumor_Sample_Barcode".

meta data.table object containing meta data.

MutationAnnotationFormat_v2.0-class

Class MutationAnnotationFormat_v2.0

Description

An S4 class to represent data in mutation annotation format version 2.0, inherits from the MutationAnnotationFormat_Virtual class.

Usage

```
MutationAnnotationFormat_v2.0(mafData)
```

Arguments

mafData data.table object containing a maf file conforming to the version 2.0 specification.

Slots

position data.table object containing column names "Chromosome", "Start_Position", "End_Position", "Strand".

mutation data.table object containing column names "Variant_Classification", "Variant_Type", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2".

sample data.table object containing columns names "Tumor_Sample_Barcode".

meta data.table object containing meta data.

MutationAnnotationFormat_v2.1-class

Class MutationAnnotationFormat_v2.1

Description

An S4 class to represent data in mutation annotation format version 2.1, inherits from the MutationAnnotationFormat_Virtual class.

Usage

MutationAnnotationFormat_v2.1(mafData)

Arguments

mafData data.table object containing a maf file conforming to the version 2.1 specification.

Slots

position data.table object containing column names "Chromosome", "Start_Position", "End_Position", "Strand".

mutation data.table object containing column names "Variant_Classification", "Variant_Type", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2".

sample data.table object containing columns names "Tumor_Sample_Barcode".

meta data.table object containing meta data.

MutationAnnotationFormat_v2.2-class

Class MutationAnnotationFormat_v2.2

Description

An S4 class to represent data in mutation annotation format version 2.2, inherits from the MutationAnnotationFormat_Virtual class.

Usage

MutationAnnotationFormat_v2.2(mafData)

Arguments

mafData data.table object containing a maf file conforming to the version 2.2 specification.

Slots

`position` data.table object containing column names "Chromosome", "Start_Position", "End_Position", "Strand".

`mutation` data.table object containing column names "Variant_Classification", "Variant_Type", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2".

`sample` data.table object containing columns names "Tumor_Sample_Barcode".

`meta` data.table object containing meta data.

MutationAnnotationFormat_v2.3-class

Class MutationAnnotationFormat_v2.3

Description

An S4 class to represent data in mutation annotation format version 2.3, inherits from the `MutationAnnotationFormat_Virtual` class.

Usage

`MutationAnnotationFormat_v2.3(mafData)`

Arguments

`mafData` data.table object containing a maf file conforming to the version 2.3 specification.

Slots

`position` data.table object containing column names "Chromosome", "Start_Position", "End_Position", "Strand".

`mutation` data.table object containing column names "Variant_Classification", "Variant_Type", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2".

`sample` data.table object containing columns names "Tumor_Sample_Barcode".

`meta` data.table object containing meta data.

MutationAnnotationFormat_v2.4-class

Class MutationAnnotationFormat_v2.4

Description

An S4 class to represent data in mutation annotation format version 2.4, inherits from the `MutationAnnotationFormat_Virtual` class.

Usage

`MutationAnnotationFormat_v2.4(mafData)`

Arguments

mafData data.table object containing a maf file conforming to the version 2.4 specification.

Slots

position data.table object containing column names "Chromosome", "Start_Position", "End_Position", "Strand".

mutation data.table object containing column names "Variant_Classification", "Variant_Type", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2".

sample data.table object containing columns names "Tumor_Sample_Barcode".

meta data.table object containing meta data.

MutationAnnotationFormat_Virtual-class

Class MutationAnnotationFormat_Virtual

Description

An S4 class to act as a virtual class for MutationAnnotationFormat version sub-classes.

Slots

position data.table object holding genomic positions.

mutation data.table object holding mutation status data.

sample data.table object holding sample data.

meta data.table object holding all other meta data.

MutSpectra-class

Class MutSpectra

Description

An S4 class for the MutSpectra plot object, under development!!!

Usage

```
MutSpectra(object, BSgenome = NULL, sorting = NULL, palette = NULL,
  clinical = NULL, sectionHeights = NULL, sampleNames = TRUE,
  verbose = FALSE, plotALayers = NULL, plotBLayers = NULL,
  plotCLayers = NULL)
```

Arguments

| | |
|----------------|---|
| object | Object of class MutationAnnotationFormat, GMS, VEP. |
| BSgenome | Object of class BSgenome, used to extract reference bases if not supplied by the file format. |
| sorting | Character vector specifying how samples should be ordered in the plot, one of "mutation", "sample", or a vector of length equal to the number of samples explicitly providing the order of samples. |
| palette | Character vector specifying the colors used for encoding transitions and transversions, should be of length 6. If NULL a default palette will be used. |
| clinical | Object of class Clinical, used for adding a clinical data subplot. |
| sectionHeights | Numeric vector specifying relative heights of each plot section, should sum to one. Expects a value for each section. |
| sampleNames | Boolean specifying if samples should be labeled on the plot. |
| verbose | Boolean specifying if status messages should be reported |
| plotALayers | list of ggplot2 layers to be passed to the frequency plot. |
| plotBLayers | list of ggplot2 layers to be passed to the proportion plot. |
| plotCLayers | list of ggplot2 layers to be passed to the clinical plot. |

Slots

| | |
|--------------|---|
| PlotA | gtable object for the mutation frequencies. |
| PlotB | gtable object for the mutation proportions. |
| PlotC | gtable object for clinical data sub-plot. |
| Grob | gtable object for the arranged plot. |
| primaryData | data.table object storing the primary data, should have column names sample, mutation, frequency, proportion. |
| ClinicalData | data.table object storing the data used to plot the clinical sub-plot. |

 PIK3CA

Subset MAF file for PIK3CA gene

Description

A data set originating from the open access TCGA data (6c93f518-1956-4435-9806-37185266d248), the data set is composed of mutations for the PIK3CA gene for breast cancer. this is primarily intended to test the Lollipop() function.

Usage

```
data(PIK3CA)
```

Format

a data frame with 361 observations and 19 variables

Value

Object of class data frame

| | |
|----------------|-----------------------|
| Rainfall-class | <i>Class Rainfall</i> |
|----------------|-----------------------|

Description

An S4 class for the Rainfall plot object, under development!!!

Usage

```
Rainfall(object, BSgenome = NULL, palette = NULL,
          sectionHeights = NULL, chromosomes = NULL, sample = NULL,
          pointSize = NULL, verbose = FALSE, plotALayers = NULL,
          plotBLayers = NULL)
```

Arguments

| | |
|----------------|---|
| object | Object of class MutationAnnotationFormat, GMS, VEP. |
| BSgenome | Object of class BSgenome to extract genome wide chromosome coordinates |
| palette | Character vector specifying colors used for encoding transitions and transversions , should be of length 7. If NULL a default palette will be used. |
| sectionHeights | Numeric vector specifying relative heights of each plot section, should sum to one. Expects a value for each section. |
| chromosomes | Character vector specifying chromosomes for which to plot |
| sample | Character vector specifying the samples for which to plot. |
| pointSize | numeric value giving the size of points to plot (defaults to 2) |
| verbose | Boolean specifying if status messages should be reported. |
| plotALayers | list of ggplot2 layers to be passed to the rainfall plot. |
| plotBLayers | list of ggplot2 layers to be passed to the density plot. |

Slots

PlotA gtable object for the rainfall plot

PlotB gtable object for density plots based on the rainfall plot

Grob gtable object for the arranged plot

primaryData data.table object storing the primary data used for plotting.

SNPloci *Identity snps*

Description

A data set containing locations of 24 identity snps originating from: Pengelly et al. Genome Med. 2013, PMID 24070238

Usage

```
data(SNPloci)
```

Format

a data frame with 24 observations and 3 variables

Value

Object of class data frame

TvTi *Construct transition-transversion plot*

Description

Given a data frame construct a plot displaying the proportion or frequency of transition and transversion types observed in a cohort.

Usage

```
TvTi(x, fileType = NULL, y = NULL, clinData = NULL,
     type = "Proportion", lab_Xaxis = TRUE, lab_txtAngle = 45,
     palette = c("#D53E4F", "#FC8D59", "#FEE08B", "#E6F598", "#99D594",
                 "#3288BD"), tvtiLayer = NULL, expecLayer = NULL, sort = "none",
     clinLegCol = NULL, clinVarCol = NULL, clinVarOrder = NULL,
     clinLayer = NULL, progress = TRUE, out = "plot",
     sample_order_input, layers = NULL, return_plot = FALSE)
```

Arguments

| | |
|----------|---|
| x | Object of class data frame with rows representing transitions and transversions. The data frame must contain the following columns 'sample', 'reference' and 'variant' or alternatively "Tumor_Sample_Barcode", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2" depending on the argument supplied to the fileType parameter. (required) |
| fileType | Character string specifying the format the input given to parameter x is in, one of 'MAF', 'MGI'. The former option requires the data frame given to x to contain the following column names "Tumor_Sample_Barcode", "Reference_Allele", "Tumor_Seq_Allele1", "Tumor_Seq_Allele2" the later option requires the data frame givin to x to contain the following column names "reference", "variant" and "sample". (required) |

| | |
|--------------------|---|
| y | Named vector or data frame representing the expected transition and transversion rates. Either option must name transition and transversions as follows: "A->C or T->G (TV)", "A->G or T->C (TI)", "A->T or T->A (TV)", "G->A or C->T (TI)", "G->C or C->G (TV)", "G->T or C->A (TV)". If specifying a data frame, the data frame must contain the following columns names "Prop", "trans_tranv" (optional see vignette). |
| clinData | Object of class data frame with rows representing clinical data. The data frame should be in "long format" and columns must be names as "sample", "variable", and "value" (optional see details and vignette). |
| type | Character string specifying if the plot should display the Proportion or Frequency of transitions/transversions observed. One of "Proportion" or "Frequency", defaults to "Proportion". |
| lab_Xaxis | Boolean specifying whether to label the x-axis in the plot. |
| lab_txtAngle | Integer specifying the angle of labels on the x-axis of the plot. |
| palette | Character vector of length 6 specifying colours for each of the six possible transition transversion types. |
| tvtiLayer | Valid ggplot2 layer to be added to the main plot. |
| expecLayer | Valid ggplot2 layer to be added to the expected sub-plot. |
| sort | Character string specifying the sort order of the sample variables in the plot. Arguments to this parameter should be "sample", "tvti", or "none" to sort the x-axis by sample name, transition transversion frequency, or no sort respectively. |
| clinLegCol | Integer specifying the number of columns in the legend for the clinical data, only valid if argument is supplied to parameter clinData. |
| clinVarCol | Named character vector specifying the mapping of colours to variables in the variable column of the data frame supplied to clinData (ex. "variable"="colour"). |
| clinVarOrder | Character vector specifying the order in which to plot variables in the variable column of the argument given to the parameter clinData. The argument supplied to this parameter should have the same unique length and values as in the variable column of the argument supplied to parameter clinData (see vignette). |
| clinLayer | Valid ggplot2 layer to be added to the clinical sub-plot. |
| progress | Boolean specifying if progress bar should be displayed for the function. |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |
| sample_order_input | Sample orders to be used |
| layers | ggplot object to be added to proportions plot |
| return_plot | Return as ggplot object? Only returns main plot |

Details

TvTi is a function designed to display proportion or frequency of transitions and transversion seen in a data frame supplied to parameter x.

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
TvTi(brcaMAF, type='Frequency',
palette=c("#77C55D", "#A461B4", "#C1524B", "#93B5BB", "#4F433F", "#BFA753"),
lab_txtAngle=60, fileType="MAF")
```

| | |
|-----------|------------------|
| VEP-class | <i>Class VEP</i> |
|-----------|------------------|

Description

An S4 class for Variant Effect Predictor input, under development!!!

Usage

```
VEP(path, data = NULL, version = "auto", verbose = FALSE)
```

Arguments

| | |
|---------|---|
| path | String specifying the path to a VEP annotation file. Can accept wildcards if multiple VEP annotation files exist (see details). |
| data | data.table object storing a GMS annotation file. Overrides "path" if specified. |
| version | String specifying the version of the VEP files, Defaults to auto which will look for the version in the header. |
| verbose | Boolean specifying if progress should be reported while reading in the VEP files. |

Details

When specifying a path to a VEP annotation file the option exist to either specify the full path to an annotation file or to use wildcards to specify multiple files. When specifying a full path the initializer will check if a column named "sample" containing the relevant sample for each row exists. If such a column is not found the initializer will assume this file corresponds to only one sample and populate a sample column accordingly. Alternatively if multiple files are specified at once using a wildcard, the initializer will aggregate all the files and use the file names minus any extension to populate sample names.

Slots

path Character string specifying the paths of the VEP files read in.
version Numeric value specifying the version of VEP used.
vepObject vep object which inherits from VEP_Virtual class.

See Also

[Waterfall](#)
[MutSpectra](#)

VEP_v88-class *Class VEP_v88*

Description

An S4 class to represent data in variant effect predictor version 88 format, inherits from the VEP_Virtual class, under development!!!

Usage

```
VEP_v88(vepData, vepHeader)
```

Arguments

vepData data.table object containing a VEP annotation file conforming to the version 88 specifications.

vepHeader Object of class list containing character vectors for vep header information.

Slots

header data.table object containing header information

description data.table object containing column descriptions

position data.table object containing column names "chromosome_name", "start", "stop".

mutation data.table object containing column names "reference", "variant", "trv_type".

sample data.table object containing columns names "sample".

meta data.table object containing meta data.

VEP_Virtual-class *Class VEP_Virtual*

Description

An S4 class to act as a virtual class for VEP version sub-classes, under development!!!

Slots

header data.table object holding header information.

description data.table object holding column descriptions

position data.table object holding genomic positions.

mutation data.table object holding mutation status data.

sample data.table object holding sample data.

meta data.table object holding all other meta data.

 waterfall

Construct a waterfall plot

Description

Given a data frame construct a water fall plot showing the mutation burden and mutation type on a gene and sample level.

Usage

```
waterfall(x, mainRecurCutoff = 0, mainGrid = TRUE,
  mainXlabel = FALSE, main_geneLabSize = 8, mainLabelCol = NULL,
  mainLabelSize = 4, mainLabelAngle = 0, mainDropMut = FALSE,
  mainPalette = NULL, mainLayer = NULL, mutBurden = NULL,
  plotMutBurden = TRUE, coverageSpace = 44100000,
  mutBurdenLayer = NULL, clinData = NULL, clinLegCol = 1,
  clinVarOrder = NULL, clinVarCol = NULL, clinLayer = NULL,
  sampRecurLayer = NULL, plotGenes = NULL, geneOrder = NULL,
  plotSamples = NULL, sampOrder = NULL, maxGenes = NULL,
  rmvSilent = FALSE, fileType = "MAF", variant_class_order = NULL,
  out = "plot", plot_proportions = FALSE, proportions_layer = NULL,
  proportions_type = "TRV_TYPE", section_heights)
```

Arguments

- | | |
|------------------|--|
| x | Object of class data frame representing annotated mutations. The data frame supplied must have one of the following sets of column names ("Tumor_Sample_Barcode", "Hugo_Symbol", "Variant_Classification") for fileType="MAF", ("sample", "gene_name", "trv_type") for fileType="MGI" or ("sample", "gene", "variant_class") for fileType="Custom". This columns should represent samples in a cohort, gene with mutation, and the mutation type respectively. |
| mainRecurCutoff | Numeric value between 0 and 1 specifying a mutation recurrence cutoff. Genes which do not have mutations in the proportion os samples defined are removed. |
| mainGrid | Boolean specifying if a grid should be overlayed on the main plot. Not recommended if the number of genes or samples to be plotted is large. |
| mainXlabel | Boolean specifying whether to label the x-axis with sample names. Not recommended if the number of samples to be plotted is large. |
| main_geneLabSize | Intenger specifying the size of gene names displayed on the y-axis. |
| mainLabelCol | Character string specifying a column name from the argument supplied to parameter 'x' from which to derive cell labels from (see details and vignette). |
| mainLabelSize | Integer specifying the size of text labels for cells in the main plot. Valid only if argument is supplied to the parameter 'mainLabelCol'. |
| mainLabelAngle | Integer specifying the degree of rotation for text labels. Valid only if argument is supplied to the parameter 'mainLabelCol'. |
| mainDropMut | Boolean specifying whether to drop unused "mutation type" levels from the legend. |

| | |
|---------------------|---|
| mainPalette | Character vector specifying colours for mutation types plotted in the main plot, must specify a colour for each mutation type plotted. |
| mainLayer | Valid ggplot2 layer to be added to the main plot. |
| mutBurden | Object of class data frame containing columns "sample", "mut_burden" with sample levels matching those supplied in x. |
| plotMutBurden | Boolean specify if the mutation burden sub-plot should be displayed. |
| coverageSpace | Integer specifying the size in bp of the genome covered by sequence data from which mutations could be called (see details and vignette). |
| mutBurdenLayer | Valid ggplot2 layer to be added to the top sub-plot. |
| clinData | Object of class data frame with rows representing clinical data. The data frame should be in "long format" and columns must be names as "sample", "variable", and "value" (optional see details and vignette). |
| clinLegCol | Integer specifying the number of columns in the legend for the clinical data, only valid if argument is supplied to parameter clinData. |
| clinVarOrder | Character vector specifying the order in which to plot variables in the variable column of the argument given to the parameter clinData. The argument supplied to this parameter should have the same unique length and values as in the variable column of the argument supplied to parameter clinData (see vignette). |
| clinVarCol | Named character vector specifying the mapping of colours to variables in the variable column of the data frame supplied to clinData (ex. "variable"="colour"). |
| clinLayer | Valid ggplot2 layer to be added to the clinical sub-plot. |
| sampRecurLayer | Valid ggplot2 layer to be added to the left sub-plot. |
| plotGenes | Character vector specifying genes to plot. If not null genes not specified within this character vector are removed. |
| geneOrder | Character vector specifying the order in which to plot genes. |
| plotSamples | Character vector specifying samples to plot. If not null all other samples not specified within this parameter are removed. |
| sampOrder | Character vector specifying the order of the samples to plot. |
| maxGenes | Integer specifying the maximum number of genes to be plotted. Genes kept will be chosen based on the recurrence of mutations in samples. |
| rmvSilent | Boolean specifying if silent mutations should be removed from the plot. |
| fileType | Character string specifying the file format of the data frame specified to parameter 'x', one of "MGI", "MAF", "Custom" (see details and vignette). |
| variant_class_order | Character vector specifying the hierarchical order of mutation types to plot, required if file_type == "Custom" (see details and vignette). |
| out | Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns). |
| plot_proportions | Plot mutational profile layer? |
| proportions_layer | ggplot2 layer(s) to be added to the mutational profile plot |
| proportions_type | Which type of proportions plot to use? Can be "trv_type" or "TvTi" currently |
| section_heights | Heights of each section. Must be the same length as the number of vertical section |

Details

waterfall is a function designed to visualize the mutations seen in a cohort. The function takes a data frame with appropriate column names (see fileType parameter) and plots the mutations within. In cases where multiple mutations occur in the same cell the most deleterious mutation is given priority (see vignette for default priority). If the fileType parameter is set to "Custom" the user must supply this priority via the 'variant_class_order' parameter with the highest priorities occurring first. Additionally this parameter will override the default orders of MGI and MAF file types.

Various data subsets are allowed via the waterfall function (see above), all of these subsets will occur independently of the mutation burden calculation. To clarify the removal of genes and mutations will only occur after the mutation burden is calculated. The mutation burden calculation is only meant to provide a rough estimate and assumes that the coverage breadth within the cohort is approximately equal. For more accurate calculations it is recommended to supply this information via the mutBurden parameter which. Note that the mutation burden calculation relies on the 'coverageSpace' parameter (see vignette).

It is possible to display additional information within the plot via cell labels. The 'mainLabelCol' parameter will look for an additional column in the data frame and plot text within cells based on those values (see vignette).

Value

One of the following, a list of dataframes containing data to be plotted, a grob object, or a plot.

Examples

```
# Plot the data
waterfall(brcaMAF, plotGenes=c("PIK3CA", "TP53", "USH2A", "MLL3", "BRCA1"))
```

| | |
|-----------------|------------------------|
| Waterfall-class | <i>Class Waterfall</i> |
|-----------------|------------------------|

Description

An S4 class for the waterfall plot object, under development!!!

Usage

```
Waterfall(input, labelColumn = NULL, samples = NULL, coverage = NULL,
  mutation = NULL, genes = NULL, mutationHierarchy = NULL,
  recurrence = NULL, geneOrder = NULL, geneMax = NULL,
  sampleOrder = NULL, plotA = c("frequency", "burden", NULL),
  plotATally = c("simple", "complex"), plotALayers = NULL,
  plotB = c("proportion", "frequency", NULL), plotBTally = c("simple",
  "complex"), plotBLayers = NULL, gridOverlay = FALSE, drop = TRUE,
  labelSize = 5, labelAngle = 0, sampleNames = TRUE,
  clinical = NULL, sectionHeights = NULL, sectionWidths = NULL,
  verbose = FALSE, plotCLayers = NULL)
```

Arguments

| | |
|-------------------|---|
| input | Object of class <code>MutationAnnotationFormat</code> , <code>VEP</code> , <code>GMS</code> , or alternatively a data frame/data table with column names "sample", "gene", "mutation". |
| labelColumn | Character vector specifying a column name from which to extract label names for cells, must be a column within the object passed to input. |
| samples | Character vector specifying samples to plot. If not NULL all samples in "input" not specified with this parameter are removed. Further samples specified but not present in the data will be added. |
| coverage | Integer specifying the size in base pairs of the genome covered by sequence data from which mutations could be called. Required for the mutation burden sub-plot (see details and vignette). Optionally a named vector of integers corresponding to each sample can be supplied for more accurate calculations. |
| mutation | Character vector specifying mutations to keep, if defined mutations not supplied are removed from the main plot. |
| genes | Character vector specifying genes to keep, if not "NULL" all genes not specified are removed. Further genes specified but not present in the data will be added. |
| mutationHierarchy | data.table/data.frame object with rows specifying the order of mutations from most to least deleterious and containing column names "mutation" and "color". Used to change the default colors and/or to give priority to a mutation for the same gene/sample (see details and vignette). |
| recurrence | Numeric value between 0 and 1 specifying a mutation recurrence cutoff. Genes which do not have mutations in the proportion of samples defined are removed. |
| geneOrder | Character vector specifying the order in which to plot genes. |
| geneMax | Integer specifying the maximum number of genes to be plotted. Genes kept will be chosen based on the recurrence of mutations in samples, unless geneOrder is specified. |
| sampleOrder | Character vector specifying the order in which to plot samples. |
| plotA | String specifying the type of plot for the top sub-plot, one of "burden", "frequency", or NULL for a mutation burden (requires coverage to be specified), frequency of mutations, or no plot respectively. |
| plotATally | String specifying one of "simple" or "complex" for a simplified or complex tally of mutations respectively. |
| plotALayers | list of ggplot2 layers to be passed to the plot. |
| plotB | String specifying the type of plot for the left sub-plot, one of "proportion", "frequency", or NULL for a plot of gene proportions frequencies, or no plot respectively. |
| plotBTally | String specifying one of "simple" or "complex" for a simplified or complex tally of genes respectively. |
| plotBLayers | list of ggplot2 layers to be passed to the plot. |
| gridOverlay | Boolean specifying if a grid should be overlaid on the waterfall plot. This is not recommended for large cohorts. |
| drop | Boolean specifying if mutations not in the main plot should be dropped from the legend. If FALSE the legend will be based on mutations in the data before any subsets occur. |
| labelSize | Integer specifying the size of label text within each cell if "labelColumn" has been specified. |

| | |
|----------------|---|
| labelAngle | Numeric value specifying the angle of label text if "labelColumn" has been specified. |
| sampleNames | Boolean specifying if samples should be labeled on the x-axis of the plot. |
| clinical | Object of class Clinical , used for adding a clinical data subplot. |
| sectionHeights | Numeric vector specifying relative heights of each plot section, should sum to one. Expects a value for each section. |
| sectionWidths | Numeric vector specifying relative heights of each plot section, should sum to one. Expects a value for each section. |
| verbose | Boolean specifying if status messages should be reported. |
| plotCLayers | list of ggplot2 layers to be passed to the main plot. |

Details

‘Waterfall()’ is designed to visualize the mutations seen in a cohort. As input the function takes an object of class [MutationAnnotationFormat](#), [VEP](#), or [GMS](#). Alternatively a user can provide either of `data.table` or `data.frame` as long as the column names of those objects include "sample", "gene", and "mutation". When supplying an object of class `data.table` or `data.frame` the user must also provide input to the ‘mutationHierarchy’ parameter.

The ‘mutationHierarchy’ parameter expects either a `data.table` or `data.frame` object containing the column names "mutation" and "color". Each row should match a mutation type given in the param ‘input’. The ‘mutationHierarchy’ parameter is intended to both change the colors of mutations on the plot and to set a hierarchy of which mutation type to plot if there are more than 1 mutation types for the same gene/sample combination.

Slots

| | |
|-----------------------|---|
| PlotA | <code>gtable</code> object for the top sub-plot. |
| PlotB | <code>gtable</code> object for the left sub-plot. |
| PlotC | <code>gtable</code> object for the main plot. |
| PlotD | <code>gtable</code> object for the bottom sub-plot. |
| Grob | <code>gtable</code> object for the arranged plot. |
| primaryData | <code>data.table</code> object storing the primary data, should have column names sample, gene, mutation, label. |
| simpleMutationCounts | <code>data.table</code> object storing simplified mutation counts, should have column names sample, mutation, Freq, mutationBurden |
| complexMutationCounts | <code>data.table</code> object storing mutation counts per mutation type should have column names sample, mutation, Freq, mutationBurden. |
| geneData | <code>data.table</code> object storing gene counts, should have column names gene, mutation, count. |
| ClinicalData | <code>data.table</code> object storing the data used to plot the clinical sub-plot. |
| mutationHierarchy | <code>data.table</code> object storing the hierarchy of mutation type in order of most to least important and the mapping of mutation type to color. Should have column names mutation, color, and label. |

See Also

[MutationAnnotationFormat](#), [VEP](#), [GMS](#), [Clinical](#)

Examples

```

set.seed(426)

# create a data frame with required column names
mutationDF <- data.frame("sample"=sample(c("sample_1", "sample_2", "sample_3"), 10, replace=TRUE),
                        "gene"=sample(c("egfr", "tp53", "rb1", "apc"), 10, replace=TRUE),
                        "mutation"=sample(c("missense", "frame_shift", "splice_site"), 10, replace=TRUE))

# set the mutation hierarchy (required for DF)
hierarchyDF <- data.frame("mutation"=c("missense", "frame_shift", "slice_site"),
                        "color"=c("#3B3B98", "#BDC581", "#6A006A"))

# Run the Waterfall Plot and draw the output
Waterfall.out <- Waterfall(mutationDF, mutationHierarchy=hierarchyDF)
drawPlot(Waterfall.out)

```

writeData*Method writeData*

Description

Method writeData

Usage

```

writeData(object, ...)

## S4 method for signature 'GMS_Virtual'
writeData(object, file, sep, ...)

## S4 method for signature 'GMS'
writeData(object, file, ...)

## S4 method for signature 'MutationAnnotationFormat_Virtual'
writeData(object, file, sep,
          ...)

## S4 method for signature 'MutationAnnotationFormat'
writeData(object, file, ...)

## S4 method for signature 'VEP_Virtual'
writeData(object, file, sep, ...)

## S4 method for signature 'VEP'
writeData(object, file, ...)

```

Arguments

| | |
|--------|---|
| object | Object of class VEP |
| ... | additional arguments to passed |
| file | Character string specifying a file to send output to. |
| sep | Delimiter used when writing output, defaults to tab. |

Details

The `writeData` method is used to output data held in GenVisR objects to a file.

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