

# Package ‘GenVisR’

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**Title** Genomic Visualizations in R

**Version** 1.8.1

**Maintainer** Zachary Skidmore <zlskidmore@gmail.com>

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

**Depends** R (>= 3.3.0)

**Imports** AnnotationDbi, biomaRt, BiocGenerics, Biostrings, DBI, FField, GenomicFeatures, GenomicRanges (>= 1.25.4), ggplot2 (>= 2.1.0), grid, gridExtra, gtable, gtools, IRanges (>= 2.7.5), plyr (>= 1.8.3), reshape2, Rsamtools, scales, stats, utils, viridis

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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

**VignetteBuilder** knitr

**RoxygenNote** 6.0.1.9000

**NeedsCompilation** no

**Author** Zachary Skidmore [aut, cre], Alex Wagner [aut], Robert Lesurf [aut], Katie Campbell [aut], Jason Kunisaki [aut], Obi Griffith [aut], Malachi Griffith [aut]

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brcaMAF	<i>Truncated BRCA MAF file</i>
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### 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

---

cnFreq	<i>Construct copy-number frequency plot</i>
--------	---

---

### 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**

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

---

cnFreq_buildMain	<i>Construct CN frequency plot</i>
------------------	------------------------------------

---

**Description**

given a data frame construct a plot to display proportions of losses and gains across the genome

**Usage**

```
cnFreq_buildMain(x, plotType, dummy_data, plot_title = NULL,
  CN_low_colour = "#002EB8", CN_high_colour = "#A30000", x_lab_size = 12,
  y_lab_size = 12, facet_lab_size = 10, plotLayer = NULL)
```

**Arguments**

x	object of class data frame containing columns chromosome, start, end, gain, and loss
plotType	character string to determine whether to plot proportions or frequencies
dummy_data	Object of class data frame containing columns chromosome, start, end, cn, sample. Used for defining chromosome boundaries
plot_title	character string for title of plot
CN_low_colour	character string specifying low value of colour gradient
CN_high_colour	character string specifying high value of colour gradient
x_lab_size	integer specifying the size of the X label
y_lab_size	integer specifying the size of the Y label
facet_lab_size	integer specifying the size of the faceted labels
plotLayer	Additional layer to be plotted, can be a theme but must be a ggplot layer

**Value**

ggplot object

---

cnFreq_qual	<i>check input to cnFreq</i>
-------------	------------------------------

---

**Description**

Perform a data quality check for input to cnFreq

**Usage**

```
cnFreq_qual(x)
```

**Arguments**

x	a data frame with columns chromosome, start, end, segmean, and sample
---	---

**Value**

data frame passing quality checks

---

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 desirable 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")
```

---

cnSpec_buildMain	<i>Construct CN cohort plot</i>
------------------	---------------------------------

---

**Description**

given a data frame construct a plot to display CN information for a group of samples

**Usage**

```
cnSpec_buildMain(data_frame, dummy_data, plot_title = NULL,
  CN_low_colour = "#002EB8", CN_high_colour = "#A30000", x_lab_size = 12,
  y_lab_size = 12, facet_lab_size = 10, layers = NULL,
  CNscale = "absolute")
```

**Arguments**

data_frame	object of class data frame containing columns chromosome, start, end, cn, sample
dummy_data	Object of class data frame containing columns chromosome, start, end, cn, sample. Used for defining chromosome boundaries
plot_title	character string for title of plot
CN_low_colour	character string specifying low value of colour gradient
CN_high_colour	character string specifying high value of colour gradient
x_lab_size	integer specifying the size of the X label
y_lab_size	integer specifying the size of the Y label
facet_lab_size	integer specifying the size of the faceted labels
layers	Additional layers to be plotted, can be a theme but must be a ggplot layer
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"

**Value**

ggplot object



---

cnSpec_qual	<i>Construct CN cohort plot</i>
-------------	---------------------------------

---

**Description**

given a data frame construct a plot to display CN information for a group of samples

**Usage**

```
cnSpec_qual(x, y, genome, CNscale)
```

**Arguments**

x	object of class data frame containing columns chromosome, start, stop, seg-mean, sample
y	object of class data frame containing user supplied chromosome locations
genome	character string specifying a user supplied genome
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"

**Value**

character string specifying input passed quality check

---

cnView	<i>Construct copy-number single sample plot</i>
--------	---

---

**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).

<code>z</code>	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)
<code>genome</code>	Character string specifying a valid UCSC genome (see details).
<code>chr</code>	Character string specifying which chromosome to plot one of "chr..." or "all"
<code>CNscale</code>	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"
<code>ideogram_txtAngle</code>	Integer specifying the angle of cytogenetic labels on the ideogram subplot.
<code>ideogram_txtSize</code>	Integer specifying the size of cytogenetic labels on the ideogram subplot.
<code>plotLayer</code>	Valid ggplot2 layer to be added to the copy number plot.
<code>ideogramLayer</code>	Valid ggplot2 layer to be added to the ideogram sub-plot.
<code>out</code>	Character vector specifying the the object to output, one of "data", "grob", or "plot", defaults to "plot" (see returns).
<code>segmentColor</code>	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)
```

---

cnView_buildMain	<i>construct CN plot</i>
------------------	--------------------------

---

**Description**

given a CN data frame plot points in ggplot

**Usage**

```
cnView_buildMain(x, y, z = NULL, chr, CNscale = FALSE, layers = NULL,
  segmentColor = NULL)
```

**Arguments**

x	a data frame with columns chromosome, coordinate, cn, p_value
y	a data frame with columns chromosome, coordinate for plotting boundaries
z	a data frame with columns chromosome, start, end, segmean specifying segments called from copy number (optional)
chr	a character string specifying chromosome
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"
layers	additional ggplot2 layers to add
segmentColor	Character string specifying the color of segment lines. Used only if Z is not null.

**Value**

ggplot2 object

---

cnView_qual	<i>check input to cnView</i>
-------------	------------------------------

---

**Description**

Perform a data quality check for inputs to cnView

**Usage**

```
cnView_qual(x, y, z, genome, CNscale)
```

**Arguments**

x	a data frame with columns chromosome, coordinate, cn
y	a data frame with columns "chrom", "chromStart", "chromEnd", "name", "geneS-tain"
z	a data frame with columns chromosome, start, end, segmean
genome	character string specifying UCSC genome to use
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"

**Value**

a list of data frames passing quality checks

---

compIdent	<i>Construct identity snp comparison plot</i>
-----------	---

---

**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)
```

---

compIdent\_bamRcnt      *Count nucleotide reads at SNP locations*

---

**Description**

Given the bam file path, count the number of reads at specified snp locations

**Usage**

```
compIdent_bamRcnt(bamfile, genome, target = NULL, debug = FALSE)
```

**Arguments**

bamfile	Path to the bam file
genome	Object of class BSgenome corresponding to a genome of interest
target	Object of class data frame containing target locations in 1-base format and containing columns "chr", "start", "end", "var", "name"
debug	Boolean specifying if test datasets should be used for debugging.

**Value**

object of class data frame containing readcount information

---

compIdent\_bamRcnt\_qual      *Count nucleotide reads at SNP locations*

---

**Description**

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

**Usage**

```
compIdent_bamRcnt_qual(genome, targetbed)
```

**Arguments**

genome	Object of class BSgenome corresponding to a genome of interest
targetbed	Object of class data frame containing target locations in .bed format

**Value**

list of data objects passing quality checks

---

compIdent\_buildMain     *Compare sample identities*

---

**Description**

Produce an identity SNP plot displaying VAFs of 24 SNP locations and coverage information to compare multiple sample identities

**Usage**

```
compIdent_buildMain(x, mainLayer = NULL, covLayer = NULL)
```

**Arguments**

x	Data frame of vaf for each sample
mainLayer	Valid ggplot2 layer for altering the main plot.
covLayer	Valid ggplot2 layer for altering the coverage plot.

**Value**

ggplot2 grob object

---

compIdent\_format     *Format readcount tables from compIdent*

---

**Description**

Format readcount tables from compIdent for input into compIdent\_buildMain

**Usage**

```
compIdent_format(x)
```

**Arguments**

x	Named list of data frames with rows of the data frame corresponding to target locations.
---	--

**Value**

Formatted data frame

---

covBars	<i>Construct an overall coverage cohort plot</i>
---------	--

---

### 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

x	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
colour	Character vector specifying colours to represent sequencing depth.
plot_title	Character string specifying the title to display on the plot.
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).

### 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)
```

---

covBars_buildMain	<i>Construct coverage cohort plot</i>
-------------------	---------------------------------------

---

**Description**

given a data frame construct a plot to display coverage as percentage bars for a group of samples

**Usage**

```
covBars_buildMain(data_frame, col, plot_title = NULL, x_lab_size = 12,
  y_lab_size = 12, facet_lab_size = 10, layers = NULL)
```

**Arguments**

data_frame	object of class data frame containing columns depth, sample, bp
col	vector of colors for the coverage bars
plot_title	character string for title of plot
x_lab_size	integer specifying the size of the X label
y_lab_size	integer specifying the size of the Y label
facet_lab_size	integer specifying the size of the faceted labels
layers	Additional layers to be plotted, can be a theme but must be a ggplot layer

**Value**

ggplot object

---

covBars_qual	<i>Construct coverage cohort plot</i>
--------------	---------------------------------------

---

**Description**

given a matrix construct a plot to display coverage as percentage bars for a group of samples

**Usage**

```
covBars_qual(x)
```

**Arguments**

x	object of class matrix containing rows for the coverage and columns the sample names
---	--

**Value**

a list of data frame and color vector



---

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

---

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)
```

---

genCov_alignPlot	<i>align plots on an axis</i>
------------------	-------------------------------

---

**Description**

given a list of plots, align them on plotting space

**Usage**

```
genCov_alignPlot(plot_list, axis = "both")
```

**Arguments**

plot_list	list of ggplot objects
axis	character string to specify the axis to align plotting space on, one of both, width, height

**Value**

ggplotGrob object

---

genCov\_assign\_ggplotGrob\_height  
*assign ggplotGrob height*

---

**Description**

assign height of ggplotGrob object

**Usage**

```
genCov_assign_ggplotGrob_height(x, max_height)
```

**Arguments**

x	ggplotGrob object
max_height	grob object specifying width to reassign ggplotGrob with

**Value**

ggplotGrob

---

genCov\_assign\_ggplotGrob\_width  
*assign ggplotGrob width*

---

**Description**

assign width of ggplotGrob object

**Usage**

```
genCov_assign_ggplotGrob_width(x, max_width)
```

**Arguments**

x	ggplotGrob object
max_width	grob object specifying width to reassign ggplotGrob with

**Value**

ggplotGrob

---

genCov_buildCov	<i>build coverage plot</i>
-----------------	----------------------------

---

**Description**

given data build a coverage plot to represent the data

**Usage**

```
genCov_buildCov(data_frame, x_limits = NULL, display_x_axis = TRUE,
  colour = "blue", plot_type = "point", layers = NULL)
```

**Arguments**

data_frame	an object of class data frame containing columns stop and cov
x_limits	vector giving x-axis limits for plot, inferred from data if not specified
display_x_axis	boolean specifying whether to plot x-axis labels
colour	character string specifying the color of the data in the plot
plot_type	character string specifying one of line, bar, or point for data display
layers	additional ggplot2 layers to plot

**Value**

ggplot object

---

genCov_buildTrack	<i>build label for plot</i>
-------------------	-----------------------------

---

**Description**

given a name create a label

**Usage**

```
genCov_buildTrack(name, bg_fill = "black", text_fill = "white",
  border = "black", size = 10)
```

**Arguments**

name	character string giving the name of the track
bg_fill	character string giving the colour to fill the label
text_fill	character string giving the colour to fill the text
border	character string specifying the colour to fill the border of the label
size	integer specifying the size of the text within the label

**Value**

ggplot object

genCov\_extr\_ggplotGrob\_height  
*extract ggplotGrob height*

---

**Description**

extract plot height of ggplotGrob object

**Usage**

```
genCov_extr_ggplotGrob_height(x)
```

**Arguments**

x                    ggplotGrob object

**Value**

ggplotGrob height parameters

---

genCov\_extr\_ggplotGrob\_width  
*extract ggplotGrob width*

---

**Description**

extract plot width of ggplotGrob object

**Usage**

```
genCov_extr_ggplotGrob_width(x)
```

**Arguments**

x                    ggplotGrob object

**Value**

ggplotGrob width parameters

---

genCov_qual	<i>Perform quality control on genCov data</i>
-------------	---

---

**Description**

Ensure data input into genCov is of the proper type and format

**Usage**

```
genCov_qual(x = x, txdb = txdb, gr = gr, genome = genome)
```

**Arguments**

x	named list containing data frames with columns end and cov
txdb	A TxDb object for a genome
gr	A Granges object specifying a region of interest
genome	Object of class BSgenome specifying the genome

**Value**

a list of objects passing basic quality control

---

genCov_trackViz	<i>Overlay tracks with plots</i>
-----------------	----------------------------------

---

**Description**

given a named list of plots, display them on tracks

**Usage**

```
genCov_trackViz(..., bgFill = "black", textFill = "white",
  border = "black", size = 10, axis_align = "none", widthRatio = c(1,
  10), list = TRUE)
```

**Arguments**

...	named list of ggplot2 plots
bgFill	character string giving the colour to fill the label
textFill	character string giving the colour to fill the text
border	character string specifying the colour to fill the border of the label
size	integer specifying the size of the text within the label
axis_align	character string specifying axis to align plotting space on, one of 'both', 'height', 'width', 'none'
widthRatio	vector of length 2 giving the ratio of track labels to plots
list	boolean specifying whether plots are in a named list or specified individually via ...

**Value**

ggplotGrob object

---

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, 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

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)
```

---

geneViz\_buildGene      *build gene plot*

---

### Description

given a data frame with gene feature information build the ggplot2 object

### Usage

```
geneViz_buildGene(data_frame, display_x_axis = TRUE, x_limits = NULL,
                  gene_colour = NULL, transcript_name = FALSE, transcript_name_size = 4,
                  layers = NULL)
```

### Arguments

data_frame	an object of class data frame specifying gene feature information
display_x_axis	Boolean specifying whether to display X axis coordinate values
x_limits	vector specifying x-axis limits of plot
gene_colour	character specifying colour of gene to be plotted
transcript_name	Boolean specifying whether to plot USCS transcript names
transcript_name_size	Integer specifying the size of the transcript name text
layers	additional ggplot2 layers to plot

**Value**

ggplot object

---

geneViz\_calcGC      *Calculate GC content*

---

**Description**

Calculate GC content for elements in a GRanges object

**Usage**

```
geneViz_calcGC(gr, genome)
```

**Arguments**

`gr`                    A GRanges object to calculate GC content for  
`genome`                Object of class BSgenome specifying the genome to calculate GC content from

**Value**

Object of class GRanges

---

geneViz\_cdsFromTXID      *cdsFromTXID*

---

**Description**

Return CDS coordinates as a GRanges object given transcript IDs

**Usage**

```
geneViz_cdsFromTXID(txdb, txid)
```

**Arguments**

`txdb`                    A TxDb object for a genome  
`txid`                    A list of TXIDs

**Value**

Object of class Granges

---

geneViz\_extrCDS      *Extract CDS*

---

**Description**

Extract CDS coordinates within a GRanges object given a transcription database

**Usage**

```
geneViz_extrCDS(txdb = NULL, gr = NULL, reduce = FALSE, gaps = FALSE)
```

**Arguments**

txdb	A TxDb object for a genome
gr	A Granges object specifying the region of interest
reduce	Boolean specifying whether to collapse isoforms
gaps	Boolean specifying whether to report space between CDS instead of CDS

**Value**

Object of class Granges list

---

geneViz\_extrUTR      *Extract UTR*

---

**Description**

Extract UTR coordinates within a GRanges object given a transcription database

**Usage**

```
geneViz_extrUTR(txdb = txdb, gr = gr, reduce = FALSE, gaps = FALSE)
```

**Arguments**

txdb	A TxDb object for a genome
gr	A Granges object specifying the region of interest
reduce	Boolean specifying whether to collapse isoforms
gaps	Boolean specifying whether to report space between UTR instead of UTR

**Value**

Object of class Granges list

---

geneViz\_formatCDS      *format cds*

---

**Description**

given a Granges object specifying a region of interest, format into a form recognizable by ggplot2

**Usage**

```
geneViz_formatCDS(txdb = NULL, gr = NULL, genome = NULL, reduce = FALSE)
```

**Arguments**

txdb	A TxDb object for a genome
gr	A Granges object to format
genome	Object of class BSgenome specifying the genome for GC content calculation
reduce	Boolean specifying whether to collapse isoforms in the Granges object ROI

**Value**

Object of class data frame

---

geneViz\_formatUTR      *format UTR*

---

**Description**

given a Granges object specifying a region of interest, format into a form recognizable by ggplot2

**Usage**

```
geneViz_formatUTR(txdb = NULL, gr = NULL, genome = NULL, reduce = FALSE)
```

**Arguments**

txdb	A TxDb object for a genome
gr	A Granges object to format
genome	Object of class BSgenome specifying the genome for GC content calculation
reduce	Boolean specifying whether to collapse isoforms in the Granges object ROI

**Value**

Object of class data frame

---

`geneViz_Granges2dataframe`*Convert Granges object to dataframe*

---

**Description**

Convert a Granges object with meta data GC content to a object of class data frame

**Usage**

```
geneViz_Granges2dataframe(gr)
```

**Arguments**

`gr` A Granges object to convert to data frame

**Value**

Object of class data frame

---

`geneViz_mapCoordSpace` *Map regions to transformed space*

---

**Description**

Reference a master genomic region file to map original positions to a transformed space

**Usage**

```
geneViz_mapCoordSpace(master, coord)
```

**Arguments**

`master` an object of class data frame containing columns start, end, width, type, trans\_start, trans\_end representing a master genomic region with features from isoforms merged

`coord` an object of class data frame containing columns start and end to map to transformed space

**Value**

an object of class data frame identical to coord but with extra columns for transformed coord

---

geneViz\_mapCovCoordSpace

*Map coverage track regions to transformed space*

---

### Description

Reference a master genomic region file to map original positions of a coverage track to a transformed space

### Usage

```
geneViz_mapCovCoordSpace(cov.coords, master)
```

### Arguments

cov.coords	an object of class data frame containing columns start and end to map to transformed space, with rows demarking single nucleotide coverages
master	an object of class data frame containing columns start, end, width, type, trans_start, trans_end representing a master genomic region with features from isoforms merged

### Value

an object of class data frame identical to coord but with extra columns for transformed coord

---

geneViz\_mergeRegions *Create Region Table*

---

### Description

Create a master region table by merging isoforms

### Usage

```
geneViz_mergeRegions(gene_features, gr, base, transform)
```

### Arguments

gene_features	A dataframe specifying features of a gene
gr	Granges object specifying the ROI
base	A vector of log bases to transform the data, corresponding to the elements of transform
transform	A vector of strings designating what objects to log transform

### Value

Master region table data frame

---

geneViz\_mergeTypeRegions      *Create Typed Region Table*

---

**Description**

Create a master region table by merging isoforms

**Usage**

```
geneViz_mergeTypeRegions(type.master)
```

**Arguments**

type.master      A dataframe of all elements of a certain type, such as CDS

**Value**

type.master A dataframe of merged elements of a certain type

---

geneViz\_mergeTypes      *Merge Typed Region Tables*

---

**Description**

Create a master region table by merging isoforms

**Usage**

```
geneViz_mergeTypes(master)
```

**Arguments**

master      An unsorted dataframe of CDS and UTR elements before merging

**Value**

master A sorted dataframe of merged CDS and UTR elements

---

GenVisR      *GenVisR*

---

**Description**

GenVisR

---

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)
```

---

ideoView\_buildMain     *build chromosome*

---

**Description**

given a data frame with cytogenetic band locations plot chromosome in ggplot

**Usage**

```
ideoView_buildMain(data_frame, chromosome, chr_txt_angle = chr_txt_angle,
  chr_txt_size = chr_txt_size, layers = NULL)
```

**Arguments**

data_frame	a data frame with columns chrom, chromStart, chromEnd, name, gieStain, height_min, height_max, alternate, bandcenter, text_y, arm
chromosome	character string specifying UCSC chromosome to plot one of chr... or all
chr_txt_angle	integer specifying angle of text when plotting band text
chr_txt_size	integer specifying size of text when plotting band text
layers	additional ggplot2 layers to plot

**Value**

ggplot object

---

ideoView\_formatCytobands  
*reformat cytogenetic band data frame*

---

**Description**

given a data frame of cytogenetic bands, format it for ggplot call

**Usage**

```
ideoView_formatCytobands(data_frame, chromosome)
```

**Arguments**

data_frame	a data frame retrieved from UCSC giving cytogenetic information
chromosome	character string specifying chromosome of interest from UCSC data frame

**Value**

object of class data frame

---

ideoView_qual	<i>Check input to ideoView</i>
---------------	--------------------------------

---

**Description**

Check that input to ideoView is properly formatted

**Usage**

```
ideoView_qual(x)
```

**Arguments**

x	a data frame with rows representing cytogenetic bands for a genome. The data frame should have columns "chrom", "chromStart", "chromEnd", "name", "gi-eStain".
---	--

**Value**

data frame

---

lohSpec	<i>Plot LOH data</i>
---------	----------------------

---

**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 afore mentioned 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)
```

---

lohSpec_buildMain	<i>Plot LOH data</i>
-------------------	----------------------

---

**Description**

Build a ggplot2 object displaying calculated LOH data

**Usage**

```
lohSpec_buildMain(x, dummyData, colourScheme = "inferno", plotLayer = NULL)
```

**Arguments**

x	object of class dataframe with loh difference calculations and column names "window_start", "window_stop", "chromosome", "sample", and "loh_diff"
dummyData	object of class dataframe with column names "chromosome", "start", "end" specifying chromosome boundaries
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. for the legend's parameters

**Value**

object of class ggplot2

---

lohSpec_fileGlob	<i>Grab data for lohSpec</i>
------------------	------------------------------

---

**Description**

Look in the specified file path and grab data with the proper extension for lohSpec

**Usage**

```
lohSpec_fileGlob(path, fileExt, step, window_size, gender)
```

**Arguments**

path	character string specifying which directory contains the sample information stored as datasets with columns "chromosome", "position", "n_vaf", "t_vaf", and "sample" (required if x is not specified)
fileExt	character string specifying the file extensions of files
step	integer with the length of divisions (bp) in chromosomes
window_size	Integer value specifying the size of the window in base pairs in which to calculate the mean Loss of Heterozygosity.
gender	vector of length equal to the number of samples, consisting of elements from the set "M", "F"

**Value**

object of class data frame from data specified in path for lohSpec

---

lohSpec_lohCalc	<i>Calculate loh difference</i>
-----------------	---------------------------------

---

**Description**

Obtain LOH on an entire chromosomes from samples in a cohort

**Usage**

```
lohSpec_lohCalc(window_data, out, normal)
```

**Arguments**

window_data	object of class data frame with columns 'window_start' and 'window_stop'
out	object of class dataframe with columns 'chromosome', 'position', 'n_vaf', 't_vaf', and 'sample'
normal	integer specifying the subtraction value from tumor VAF

**Value**

object of class dataframe containing mean LOH difference calculations and column names "window\_start", "window\_stop", "chromosome", "position", "n\_vaf", "t\_vaf", "sample", "loh\_diff"

---

lohSpec_qual	<i>Check input to lohSpec</i>
--------------	-------------------------------

---

**Description**

Perform data quality checks on input supplied to lohSpec

**Usage**

```
lohSpec_qual(x, y, genome)
```

**Arguments**

x	object of class data frame with columns 'chromosome', 'position', 'n_vaf', 't_vaf', 'sample'
y	object of class data frame with columns 'chromosome', 'start', 'end' specifying chromosomal boundaries for a genome assembly (required if genome is not specified)
genome	character string specifying the genome assembly from which input data is based

**Value**

list of inputs passing basic quality controls

---

lohSpec\_slidingWindow *Obtain LOH data*

---

### Description

Obtain LOH heatmap on entire chromosomes from samples in a cohort

### Usage

```
lohSpec_slidingWindow(loh_data, step, window_size, normal)
```

### Arguments

loh_data	data frame with columns "chromosome", "position", "n_vaf", "t_vaf", "sample" giving raw vaf calls for germline variants
step	integer with the length of divisions (bp) in chromosomes
window_size	integer with the size of the sliding window (bp) to be applied
normal	integer specifying the normal VAF frequency used in LOH calculations

### Value

object of class dataframe containing LOH data

---

lohSpec\_stepCalc *Obtain average loh within each step*

---

### Description

Calculate average LOH within each step

### Usage

```
lohSpec_stepCalc(final_dataset, step)
```

### Arguments

final_dataset	object of class dataframe with columns 'window_start', 'window_stop', 'chromosome', 'position', 'n_vaf', 't_vaf', 'sample', and 'loh_diff_avg'
step	integer with the length of divisions (bp) in chromosomes

### Value

list containing avg loh calculations for each step interval



---

lohSpec_tileCalc	<i>Calculate loh difference</i>
------------------	---------------------------------

---

**Description**

Obtain LOH on an entire chromosomes from samples in a cohort

**Usage**

```
lohSpec_tileCalc(window_data, normal)
```

**Arguments**

window_data	object of class data frame with columns "chromosome", "position", "n_vaf", "t_vaf", "sample", "bin", "window_start", "window_stop"
normal	integer specifying the subtraction value from tumor VAF

**Value**

object of class dataframe containing mean LOH difference calculations and column names "window\_start", "window\_stop", "chromosome", "position", "n\_vaf", "t\_vaf", "sample", "loh\_diff"

---

lohSpec_tilePosition	<i>Obtain window information</i>
----------------------	----------------------------------

---

**Description**

Calculate window positions to perform LOH calculation

**Usage**

```
lohSpec_tilePosition(out, window_size)
```

**Arguments**

out	object of class dataframe with columns 'chromosome', 'position', 'n_vaf', 't_vaf', and 'sample'
window_size	integer with the size of the sliding window (bp) to be applied

**Value**

list containing window start/stop positions for each chromosome from each sample to perform LOH calculations

---

lohSpec\_tileWindow      *Obtain LOH data*

---

### Description

Obtain LOH heatmap on entire chromosomes from samples in a cohort

### Usage

```
lohSpec_tileWindow(loh_data, window_size, normal)
```

### Arguments

loh_data	data frame with columns "chromosome", "position", "n_vaf", "t_vaf", "sample" giving raw vaf calls for germline variants
window_size	integer with the size of the sliding window (bp) to be applied
normal	integer specifying the normal VAF frequency used in LOH calculations

### Value

object of class dataframe containing LOH data

---

lohSpec\_windowPosition  
*Obtain window information*

---

### Description

Calculate window positions to perform LOH calculation

### Usage

```
lohSpec_windowPosition(out, step, window_size)
```

### Arguments

out	object of class dataframe with columns 'chromosome', 'position', 'n_vaf', 't_vaf', and 'sample'
step	integer with the length of divisions (bp) in chromosomes
window_size	integer with the size of the sliding window (bp) to be applied

### Value

list containing window start/stop positions for each chromosome from each sample to perform LOH calculations

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**

x	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".
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).
genome	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)
```

---

lohView_buildMain	<i>construct loh plot</i>
-------------------	---------------------------

---

**Description**

given a loh data frame plot points in ggplot

**Usage**

```
lohView_buildMain(x, y, chr, layers = NULL)
```

**Arguments**

x	a data frame with columns chromosome, position, n_vaf, t_vaf, sample
y	a data frame with columns chromosome, coordinate for plotting chromosome boundaries
chr	a character string specifying chromosome
layers	additional ggplot2 layers to add

**Value**

ggplot2 object

---

lohView_qual	<i>check input to lohView</i>
--------------	-------------------------------

---

**Description**

Perform a data quality check for inputs to lohView

**Usage**

```
lohView_qual(x, y, genome)
```

**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".
y	a data frame with columns "chrom", "chromStart", "chromEnd", "name", "gieStain"
genome	character string specifying UCSC genome to use

**Value**

a list of data frames passing quality checks

lollipop

*Construct a lollipop***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 lolli 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 lolli plotted, which were derived from the argument supplied to parameter x (see details and vignette).

<code>obsA.rep.dist.lmt</code>	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).
<code>obsA.attr.fact</code>	Numeric value representing the attraction factor for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette).
<code>obsA.adj.max</code>	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).
<code>obsA.adj.lmt</code>	Numeric value representing the adjustment limit for the lollis plotted, which were derived from the argument supplied to parameter x (see details and vignette).
<code>obsA.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 x (see details and vignette).
<code>obsB.rep.fact</code>	Numeric value representing the repulsive factor for the lollis plotted, which were derived from the argument supplied to parameter y (see details and vignette).
<code>obsB.rep.dist.lmt</code>	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 y (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 y (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 y (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 y (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 <code>ggplot2</code> 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 <code>biomaRt</code> 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\_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 specigying 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



---

lollipop_Codon2AA	<i>Convert Codon to AA</i>
-------------------	----------------------------

---

**Description**

Convert a Codon to the appropriate amino acid

**Usage**

```
lollipop_Codon2AA(x)
```

**Arguments**

x	Character string of length 1 giving the DNA codon to convert
---	--

**Value**

Character corresponding to the residue for the given codon

---

lollipop_constructGene	<i>Construct gene information</i>
------------------------	-----------------------------------

---

**Description**

Build gene for input into lollipop\_buildMain

**Usage**

```
lollipop_constructGene(gene, domain_data, length)
```

**Arguments**

gene	character string specifying gene name
domain_data	object of class data frame specifying protien domain information, obtained from lollipop_fetchDomain, should contain columns giving "description", "start", "end"
length	integer specifying length of transcript in amino acids

**Value**

object of class data frame giving gene and domain information

---

lollipop\_DNAconv      *Convert DNA character string*

---

**Description**

Convert a character string of nucleotides to amino acids or side chain class

**Usage**

```
lollipop_DNAconv(x, to = "residue")
```

**Arguments**

x	Character string of nucleotides to convert
to	Character string specifying conversion to do, one of "codon", "residue", "sidechain"

**Value**

Converted string of nucleotides as character vector

---

lollipop\_dodgeCoordX      *dodge coordinates*

---

**Description**

given amino acid position dodge on x axis

**Usage**

```
lollipop_dodgeCoordX(x, rep.fact = 5000, rep.dist.lmt = 500,
  attr.fact = 0.1, adj.max = 0.1, adj.lmt = 0.5, iter.max = 50000)
```

**Arguments**

x	numeric vector of position coordinates on x axis
rep.fact	repulsive factor for plotted mutations observed track
rep.dist.lmt	repulsive distance limit for plotted mutations observed track
attr.fact	attraction factor for plotted mutations observed track
adj.max	maximum position change for each iteration observed track
adj.lmt	position adjustment limit which simulation stops observed track
iter.max	maximum iterations beyond which to stop the simulation observed track

**Value**

numeric vector of dodged position coordinates on x axis

---

lollipop\_dodgeCoordY *dodge coordinates*

---

**Description**

given a data frame, dodge x coordinates ontop of each other

**Usage**

```
lollipop_dodgeCoordY(x, track = "top")
```

**Arguments**

x	data frame containing columns coord_x_dodge
track	character vector, one of "top", "bottom" specifying whether to dodge in a positive or negative fashion

**Value**

numeric vector of dodged position coordinates on y axis

---

lollipop\_fetchDomain *fetch protein domains*

---

**Description**

Retrieve protein domains given ensembl transcript ID

**Usage**

```
lollipop_fetchDomain(transcriptID, species = "hsapiens",  
  host = "www.ensembl.org")
```

**Arguments**

transcriptID	String specifying ensembl transcript id
species	character string to use when searching for ensemblMart dataset
host	Host to connect to.

**Value**

data frame of protien domains and start/stop coordinates

---

`lollipop_mutationObs` *format mutation observations*

---

### Description

Create a data frame of mutation observations

### Usage

```
lollipop_mutationObs(x, track, fill_value, label_column, rep.fact,
  rep.dist.lmt, attr.fact, adj.max, adj.lmt, iter.max)
```

### Arguments

<code>x</code>	object of class data frame with columns <code>trv_type</code> and amino acid change
<code>track</code>	character string specifying one to 'top', 'bottom' to specify proper track
<code>fill_value</code>	character string giving the name of the column to shade variants on
<code>label_column</code>	character string specifying column containing text information to be plotted
<code>rep.fact</code>	repulsive factor for plotted mutations observed track
<code>rep.dist.lmt</code>	repulsive distance limit for plotted mutations observed track
<code>attr.fact</code>	attraction factor for plotted mutations observed track
<code>adj.max</code>	maximum position change for each iteration observed track
<code>adj.lmt</code>	position adjustment limit which simulation stops observed track
<code>iter.max</code>	maximum iterations beyond which to stop the simulation observed track

### Value

object of class data frame giving mutation observations

---

`lollipop_qual` *Check input to lollipop*

---

### Description

Perform Basic quality checks for lollipop input

### Usage

```
lollipop_qual(x, y, z)
```

### Arguments

<code>x</code>	object of class data frame containing columns <code>transcript_name</code> , <code>gene</code> , and <code>amino_acid_change</code> and rows denoting mutations
<code>y</code>	object of class data frame containing columns <code>transcript_name</code> , and <code>amino_acid_change</code> and rows denoting mutations
<code>z</code>	Object of class data frame containing columns "description", "start", "stop" specifying gene regions to highlight

**Value**

objects passing basic quality checks

---

lollipop\_reduceLolli *Reduce Lolli*

---

**Description**

Reduce lolli stacked ontop of each other to the amount specified

**Usage**

```
lollipop_reduceLolli(x, max = NULL)
```

**Arguments**

x	Data frame with column name mutation_coord to reduce lolli on
max	Integer specifying the maximum number of lolli to allow

**Value**

Object of class data frame taking the reduced form of x

---

lollipop\_transcriptID2codingSeq  
*fetch protein length*

---

**Description**

Retrieve protein length from ensembl database given ensembl transcript id

**Usage**

```
lollipop_transcriptID2codingSeq(transcriptID, species = "hsapiens",  
  host = "www.ensembl.org")
```

**Arguments**

transcriptID	character string giving ensembl transcript id
species	character string to use when searching for ensemblMart dataset
host	Host to connect to.

**Value**

length in residues of ensembl transcript id

---

LucCNseg	<i>Truncated CN segments</i>
----------	------------------------------

---

**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

---

multi_align	<i>align CN/LOH plots on x axis</i>
-------------	-------------------------------------

---

**Description**

given a chromosome and CN/LOH plot align plot widths

**Usage**

```
multi_align(p1, p2)
```

**Arguments**

p1	ggplot object of chromosome
p2	ggplot object of CN or LOH

**Value**

ggplot object

---

multi_buildClin	<i>plot clinical information</i>
-----------------	----------------------------------

---

**Description**

given a data frame with columns names sample, variable, and value create a ggplot2 object

**Usage**

```
multi_buildClin(x, clin.legend.col = 1, clin.var.colour = NULL,  
               clin.var.order = NULL, clin.layers = NULL)
```

**Arguments**

x	a data frame in "long" format giving additional information to be plotted, requires columns "sample", "variable", and "value"
clin.legend.col	an integer specifying the number of columns to plot in the legend
clin.var.colour	a named character vector specifying the mapping between colors and variables
clin.var.order	a character vector of variables to order the legend by
clin.layers	additional ggplot2 layers to plot

**Value**

a grob object

---

multi_chrBound	<i>retrieve and format CN_cohort plot supplemental data</i>
----------------	---

---

**Description**

given a genome obtain Start and Stop positions for all chromosomes in the genome

**Usage**

```
multi_chrBound(x)
```

**Arguments**

x	data frame containing columns chromosome, start, end
---	--

**Value**

object of class data frame formatted to internal specifications

---

multi_cytobandRet	<i>Retrieve cytogenetic bands</i>
-------------------	-----------------------------------

---

**Description**

given a genome query UCSC for cytogenetic band locations

**Usage**

```
multi_cytobandRet(genome)
```

**Arguments**

genome	character string giving a UCSC genome
--------	---------------------------------------

**Value**

object of class data frame

---

multi_selectOut	<i>Choose output</i>
-----------------	----------------------

---

**Description**

Selector for choosing output for GenVisR functions

**Usage**

```
multi_selectOut(data, plot, out = "plot", draw = "FALSE")
```

**Arguments**

data	Data object to output
plot	Plot object to output
out	Character vector specifying the the object to output, one of "data", "grob", or "plot".
draw	Boolean specifying if the input to plot needs to be drawn

**Value**

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



---

multi_subsetChr	<i>subset based on chr</i>
-----------------	----------------------------

---

**Description**

given a data frame subset out specific a chromosome

**Usage**

```
multi_subsetChr(x, chr)
```

**Arguments**

x	a data frame with columns chromosome
chr	character string specifying UCSC chromosome to subset on

**Value**

object of class data frame

---

SNPloci	<i>Identity snps</i>
---------	----------------------

---

**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 given 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")
```

---

TvTi_alignPlot	<i>align TvTi plots on y axis</i>
----------------	-----------------------------------

---

## Description

align transition/transversion plots

## Usage

```
TvTi_alignPlot(p1 = NULL, p2 = NULL, p3 = NULL)
```

**Arguments**

p1            main plot  
 p2            left expected value subplot  
 p3            bottom clinical subplot

**Value**

ggplot object

---

TvTi\_annoTransTranv    *Annotate Transitions and Transversions*

---

**Description**

Given a data frame with columns reference and variant annotate the base change occurring

**Usage**

TvTi\_annoTransTranv(x)

**Arguments**

x            Object of class data frame containing columns 'reference', 'variant'

**Value**

Object of class data frame with transition/transversion annotations appended

---

TvTi\_buildMain        *build transitions/transversions*

---

**Description**

Given a data frame with columns 'trans\_tranv', 'sample', 'Freq', and 'Prop', build a transition/transversion plot

**Usage**

```
TvTi_buildMain(x, y = NULL, type = "Proportion", label_x_axis = TRUE,
  x_axis_text_angle = 45, palette = c("#D53E4F", "#FC8D59", "#FEE08B",
  "#E6F598", "#99D594", "#3288BD"), plot_expected = FALSE,
  tvti.layers = NULL, expec.layers = NULL, title_x_axis = TRUE)
```

**Arguments**

x	Object of class data frame containing columns 'trans_tranv', 'sample', 'Freq', and 'Prop'
y	Object of class data frame containing columns 'Prop', 'trans_tranv' for display of expected results
type	Object of class character specifying whether to plot the Proportion or Frequency, one of "Prop"
label_x_axis	boolean specifying wheter to label x axis
x_axis_text_angle	Integer specifying the angle to labels on x_axis
palette	Character vector of length 6 specifying colors for trans/tranv type
plot_expected	Boolean specifying if this is the main TvTi plot or a sub plot for expected values
tvti.layers	Additional ggplot2 layers for the main plot
expct.layers	Additional ggplot2 layers for the expected values plot
title_x_axis	boolean specifying whether to display an x axis title

**Value**

GGplot Object

---

TvTi\_calcTransTranvFreq
*Calculate Transition/Transversion Frequency***Description**

Given a data frame with columns reference, variant, sample, and trans/tranv calculate the frequencies of transitions and transversion occurring.

**Usage**

```
TvTi_calcTransTranvFreq(x)
```

**Arguments**

x	Object of class data frame containing columns 'reference', 'variant', 'sample', 'trans_tranv'
---	---

**Value**

Object of class data frame with Frequency and Proportion of Transitions/Transversions appended on a sample level

---

TvTi_convMAF	<i>Convert .maf format to internal format</i>
--------------	---

---

**Description**

Convert data frame in .maf format to an internally recognized format

**Usage**

```
TvTi_convMaf(x)
```

**Arguments**

x	Object of class data frame containing columns 'Tumor_Sample_Barcode', 'Reference_Allele', 'Tumor_Seq_Allele1', 'Tumor_Seq_Allele2'
---	--

**Value**

a data frame, with column names 'sample', 'reference', 'variant'

---

TvTi_qual	<i>Check input to TvTi</i>
-----------	----------------------------

---

**Description**

Perform quality check for input to function TvTi

**Usage**

```
TvTi_qual(x, y = NULL, z = NULL, file_type = "MAF")
```

**Arguments**

x	Object of class data frame containing columns 'sample', 'reference', 'variant' for 'MGI' file or 'Tumor_Sample_Barcode', 'Reference_Allele', 'Tumor_Seq_Allele1', 'Tumor_Seq_Allele2' for 'MAF' file
y	Object of class data frame containing columns "Prop", "trans_tranv"
z	Object of class data frame containing columns "sample", "variable", "value" denoting clinical information
file_type	Character string specifying the input file type expected

**Value**

a data frame, or list of data frames passing quality checks

---

TvTi_rmIndel	<i>Remove indels</i>
--------------	----------------------

---

**Description**

Given a data frame with columns reference and variants remove all indels from data

**Usage**

```
TvTi_rmIndel(x)
```

**Arguments**

x                      Object of class data frame containing columns 'reference', 'variant'

**Value**

Object of class data frame with indels removed

---

TvTi_rmMnuc	<i>Remove multinucleotide codes</i>
-------------	-------------------------------------

---

**Description**

Given a data frame with columns reference and variants remove all multinucleotides from data

**Usage**

```
TvTi_rmMnuc(x)
```

**Arguments**

x                      Object of class data frame containing columns 'reference', 'variant'

**Value**

Object of class data frame with multi nucleotide codes removed

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_align	<i>align plots</i>
-----------------	--------------------

---

**Description**

align mutation landscape, mutation burden on sample, and mutation burden on gene plots

**Usage**

```
waterfall_align(genes, heatmap, burden, clinical, proportions, section_heights)
```

**Arguments**

genes	ggplot object displaying mutation burden on gene
heatmap	ggplot object displaying a mutation landscape
burden	ggplot object displaying mutation burden on sample
clinical	ggplot object displaying clinical information "optional"
proportions	ggplot object displaying proportion of mutation types "optional"
section_heights	Heights of each section (should sum to one)

**Value**

a grob object

---

waterfall\_buildGenePrevalance  
*plot mutation recurrence in genes*

---

**Description**

plot a bar graph displaying the percentage of samples with a mutation

**Usage**

```
waterfall_buildGenePrevalance(data_frame, gene_label_size = 8,
                               layers = NULL)
```

**Arguments**

data_frame	a data frame in MAF format
gene_label_size	numeric value indicating the size of the gene labels on the y-axis
layers	additional ggplot2 layers

**Value**

a ggplot object

---

waterfall\_buildMain *Plot a mutation heatmap*

---

**Description**

Plot a Mutation Landscape with variables sample, gene, mutation

**Usage**

```
waterfall_buildMain(data_frame, grid = TRUE, label_x = FALSE,
                    file_type = "MGI", drop_mutation = FALSE, plot_x_title = TRUE,
                    plot_label = FALSE, plot_label_size = 4, plot_palette = NULL,
                    layers = NULL, plot_label_angle = 0)
```

**Arguments**

data_frame	a data frame in MAF format
grid	boolean value whether to overlay a grid on the plot
label_x	boolean value whether to label the x axis
file_type	character string specifying the file type, one of 'MAF' or 'MGI'
drop_mutation	Boolean specifying whether to drop unused "mutation type" levels from the legend
plot_x_title	Boolean specifying whether to plot the x_axis title

plot\_label Boolean specifying whether to plot text inside each cell  
 plot\_label\_size Integer specifying text size of cell labels  
 plot\_palette Character vector specifying colors to fill on mutation type  
 layers additional ggplot2 layers to plot  
 plot\_label\_angle angle at which to plot label text if plot\_label is true

**Value**

a ggplot2 object

---

waterfall\_buildMutBurden\_A  
*plot mutation burden*

---

**Description**

plot a barchart showing mutations per MB

**Usage**

waterfall\_buildMutBurden\_A(x, coverage\_space, layers = NULL)

**Arguments**

x a data frame in MAF format  
 coverage\_space an integer specifying the coverage space in base pairs from which a mutation could occur  
 layers Additional ggplot2 layers to plot

**Value**

a ggplot object

---

waterfall\_buildMutBurden\_B  
*plot mutation burden*

---

**Description**

plot a barchart showing mutation burden given by data frame

**Usage**

waterfall\_buildMutBurden\_B(x, layers = NULL)

**Arguments**

x                    a data frame containing columns sample, mut\_burden  
 layers                additional ggplot2 layers to plot

**Value**

a ggplot object

---

waterfall\_build\_proportions  
*Build mutational profile plot*

---

**Description**

Builds a ggplot object showing individuals' mutational profile

**Usage**

```
waterfall_build_proportions(data_frame, plot_palette, file_type, layers,
                             x_label)
```

**Arguments**

data\_frame        input data.frame  
 plot\_palette     Color palette to use  
 file\_type        MAF, etc  
 layers            layer(s) to add to this plot object  
 x\_label          label this plot?

**Value**

a ggplot object

---

waterfall\_calcMutFreq *Calculate Synonymous/Nonsynonymous mutation frequency*

---

**Description**

Creates a data frame giving synonymous/nonsynonymous counts on a sample level

**Usage**

```
waterfall_calcMutFreq(x)
```

**Arguments**

x                    data frame in long format with columns sample, trv\_type

**Value**

a data frame with synonymous/nonsynonymous counts appended

---

waterfall\_Custom2anno *Convert Custom File*

---

### Description

Convert columns of a Custom annotation file into a format recognizable by internal functions

### Usage

```
waterfall_Custom2anno(x, label_col)
```

### Arguments

x                    a data frame with columns having values for sample, gene, mutation type  
 label\_col            Character string specifying the column name of a label column (optional)

### Value

a data frame coerced from custom to annotation format

---

waterfall\_geneAlt        *mutation sample cutoff gene based*

---

### Description

Subset a internal mutSpec file keeping only samples within the specified gene list

### Usage

```
waterfall_geneAlt(x, genes)
```

### Arguments

x                    a data frame in long format with columns 'gene', 'trv\_type'  
 genes                character vector listing genes to plot

### Value

a subset data frame

---

`waterfall_geneRecurCutoff`*Mutation Recurrence Cutoff*

---

**Description**

Subset a MAF file keeping only samples that meet a mutation recurrence cutoff

**Usage**

```
waterfall_geneRecurCutoff(x, recurrence_cutoff)
```

**Arguments**

`x` data frame in long format with columns 'gene', 'trv\_type', 'sample'  
`recurrence_cutoff` integer specifying removal of entries not seen in at least "x" percent of samples

**Value**

a subset data frame

---

`waterfall_geneSort`     *sort waterfall file by gene*

---

**Description**

order a waterfall file ranking genes with more mutations higher if a gene order is unspecified.

**Usage**

```
waterfall_geneSort(x, geneOrder = NULL)
```

**Arguments**

`x` Data frame with columns names "gene", "trv\_type".  
`geneOrder` Character vector specifying the order in which to plot genes.

**Value**

Character vector of ordered genes

---

waterfall\_hierarchyTRV

*Hierarchical removal of MAF entries*

---

### Description

Remove MAF entries with the same gene/sample in an ordered fashion such that the most deleterious are retained

### Usage

```
waterfall_hierarchyTRV(x, file_type, variant_class_order)
```

### Arguments

x                    a data frame in long format with columns sample, gene, trv\_type  
file\_type            The type of file to act on one of 'MAF', 'MGI', 'Custom'  
variant\_class\_order            character vector giving the hierarchical order of mutation types to plot

### Value

a data frame with multiple mutations in the same sample/gene collapsed on the most deleterious

---

waterfall\_MAF2anno

*Convert MAF File*

---

### Description

Convert columns of a mutation annotation file "MAF" into a format recognizable by internal functions

### Usage

```
waterfall_MAF2anno(x, label_col)
```

### Arguments

x                    a data frame in MAF format  
label\_col            Character string specifying the column name of a label column

### Value

a data frame coerced from MAF to TGI format



---

waterfall_MGI2anno	<i>Convert MGI File</i>
--------------------	-------------------------

---

**Description**

Convert columns of a mutation annotation file "MGI" into a format recognizable by internal functions

**Usage**

```
waterfall_MGI2anno(x, label_col)
```

**Arguments**

x	a data frame in MGI internal format
label_col	Character string specifying the column name of a label column

**Value**

a data frame coerced from MGI to internal annotation format

---

waterfall_NA2gene	<i>Assign NA samples a gene</i>
-------------------	---------------------------------

---

**Description**

Replace NA values in a gene column with the top gene name

**Usage**

```
waterfall_NA2gene(x)
```

**Arguments**

x	a data frame in anno format
---	-----------------------------

**Value**

a data frame with NA values in a gene column coerced to the top gene name

---

waterfall\_palette\_names  
*waterfall\_palette\_names*

---

**Description**

Make labels and breaks for palettes

**Usage**

```
waterfall_palette_names(palette, file_type, data_frame)
```

**Arguments**

palette	Named colour vector as input
file_type	Which file type is involved?
data_frame	Only used if file_type is "custom"

**Details**

waterfall\_palette\_names

**Value**

a named list of "breaks" and "labels"

---

waterfall\_qual      *Check input to mutSpec*

---

**Description**

Perform a data quality check on input to mutSpec

**Usage**

```
waterfall_qual(x, y, z, file_type, label_col)
```

**Arguments**

x	a data frame in annotation format
y	a data frame containing clinical data or a null object
z	a data frame containing mutation burden information or a null object
file_type	Character string specifying the input format to expect in x
label_col	Character string specifying the column name of a label column

**Value**

a list of data frames passing quality checks

---

waterfall\_rmvSilent     *Silent Mutation Removal*

---

**Description**

Subset a MAF file setting keeping only sample information if a mutation is silent

**Usage**

```
waterfall_rmvSilent(x)
```

**Arguments**

x                    a data frame with columns 'sample', 'gene', 'trv\_type'

**Value**

a subset data frame

---

waterfall\_sampAlt     *mutation sample subset sample based*

---

**Description**

Alter a mutSpec input file keeping/adding entries in a selection of samples

**Usage**

```
waterfall_sampAlt(x, samples)
```

**Arguments**

x                    a data frame in long format with columns 'sample', 'trv\_type'

samples            character vector giving samples to plot

**Value**

a subset data frame

waterfall\_sampSort     *sort samples in an internal waterfall file.*

---

**Description**

perform a hierarchical sort on samples based on the presence of mutations in an ordered list of genes if a sample order is unspecified.

**Usage**

```
waterfall_sampSort(x, sampOrder = NULL)
```

**Arguments**

x                     a data frame in long format with column names "sample", "gene", "trv\_type"  
sampOrder            Character vector specifying the order of samples to plot.

**Value**

a vector of samples in a sorted order

---

waterfall\_select\_palette  
*Helper function to select a colour palette*

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**Description**

waterfall\_select\_palette

**Usage**

```
waterfall_select_palette(file_type, custom_palette = NULL)
```

**Arguments**

file\_type            Which file type is used?  
custom\_palette      Nullable custom colour palette

**Value**

A vector of colours to be used as palette

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