

# Package ‘maftools’

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**Type** Package

**Title** Summarize, Analyze and Visualize MAF Files

**Version** 2.17.0

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**Description** Analyze and visualize Mutation Annotation Format (MAF) files from large scale sequencing studies. This package provides various functions to perform most commonly used analyses in cancer genomics and to create feature rich customizable visualizations with minimal effort.

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**URL** <https://github.com/PoisonAlien/maftools>

**BugReports** <https://github.com/PoisonAlien/maftools/issues>

**Depends** R (>= 3.3)

**Imports** data.table, grDevices, methods, RColorBrewer, Rhtslib, survival, DNACopy

**Suggests** berryFunctions, Biostrings, BSgenome, BSgenome.Hsapiens.UCSC.hg19, GenomicRanges, IRanges, knitr, mclust, MultiAssayExperiment, NMF, R.utils, RaggedExperiment, rmarkdown, S4Vectors, pheatmap, curl

**LinkingTo** Rhtslib, zlibbioc

**VignetteBuilder** knitr

**biocViews** DataRepresentation, DNASeq, Visualization, DriverMutation, VariantAnnotation, FeatureExtraction, Classification, SomaticMutation, Sequencing, FunctionalGenomics, Survival

**Encoding** UTF-8

**LazyData** TRUE

**NeedsCompilation** no

**RoxygenNote** 7.1.1

**SystemRequirements** GNU make

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---

|              |                                               |
|--------------|-----------------------------------------------|
| annovarToMaf | <i>Converts annovar annotations into MAF.</i> |
|--------------|-----------------------------------------------|

---

## Description

Converts variant annotations from Annovar into a basic MAF.

## Usage

```
annovarToMaf(
  annovar,
  Center = NULL,
  refBuild = "hg19",
  tsbCol = NULL,
  table = "refGene",
  ens2hugo = TRUE,
  basename = NULL,
  sep = "\t",
  MAFobj = FALSE,
  sampleAnno = NULL
)
```

## Arguments

|            |                                                                                                                                                                                                                                                                                                                   |
|------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| annovar    | input annovar annotation file. Can be vector of multiple files.                                                                                                                                                                                                                                                   |
| Center     | Center field in MAF file will be filled with this value. Default NA.                                                                                                                                                                                                                                              |
| refBuild   | NCBI_Build field in MAF file will be filled with this value. Default hg19.                                                                                                                                                                                                                                        |
| tsbCol     | column name containing Tumor_Sample_Barcode or sample names in input file.                                                                                                                                                                                                                                        |
| table      | reference table used for gene-based annotations. Can be 'ensGene' or 'refGene'. Default 'refGene'                                                                                                                                                                                                                 |
| ens2hugo   | If 'table' is 'ensGene', setting this argument to 'TRUE' converts all ensemble IDs to hugo symbols.                                                                                                                                                                                                               |
| basename   | If provided writes resulting MAF file to an output file.                                                                                                                                                                                                                                                          |
| sep        | field separator for input file. Default tab separated.                                                                                                                                                                                                                                                            |
| MAFobj     | If TRUE, returns results as an <a href="#">MAF</a> object.                                                                                                                                                                                                                                                        |
| sampleAnno | annotations associated with each sample/Tumor_Sample_Barcode in input annovar file. If provided it will be included in MAF object. Could be a text file or a data.frame. Ideally annotation would contain clinical data, survival information and other necessary features associated with samples. Default NULL. |

## Details

Annovar is one of the most widely used Variant Annotation tools in Genomics. Annovar output is generally in a tabular format with various annotation columns. This function converts such annovar output files into MAF. This function requires that annovar was run with gene based annotation as a first operation, before including any filter or region based annotations. Please be aware that this function performs no transcript prioritization.

e.g. `table_annovar.pl example/ex1.avinput humandb/ -buildver hg19 -out myanno -remove -protocol (refGene),cytoBand,dbnsfp30a -operation (g),r,f -nastring NA`

This function mainly uses gene based annotations for processing, rest of the annotation columns from input file will be attached to the end of the resulting MAF.

## Value

MAF table.

## References

Wang, K., Li, M. & Hakonarson, H. ANNOVAR: functional annotation of genetic variants from high-throughput sequencing data. *Nucleic Acids Res* 38, e164 (2010).

## Examples

```
var.annovar <- system.file("extdata", "variants.hg19_multianno.txt", package = "maftools")
var.annovar.maf <- annovarToMaf(annovar = var.annovar, Center = 'CSI-NUS', refBuild = 'hg19',
tsbCol = 'Tumor_Sample_Barcode', table = 'ensGene')
```

---

bamreadcounts

---

*extract nucleotide counts for targeted variants from the BAM file.*


---

## Description

Given a BAM file and target loci, ‘bamreadcounts’ fetches redcounts for A, T, G, C, Ins, and Del. Function name is an homage to <https://github.com/genome/bam-readcount>

## Usage

```
bamreadcounts(
  bam = NULL,
  loci = NULL,
  zerobased = FALSE,
  mapq = 10,
  sam_flag = 1024,
  op = NULL,
  fa = NULL,
  nthreads = 4
)
```

**Arguments**

|           |                                                                                                                                                     |
|-----------|-----------------------------------------------------------------------------------------------------------------------------------------------------|
| bam       | Input bam file(s). Required.                                                                                                                        |
| loci      | Loci file. Can be a tsv file or a data.frame. First two columns should contain chromosome and position (by default assumes coordinates are 1-based) |
| zerobased | are coordinates zero-based. Default FALSE.                                                                                                          |
| mapq      | Map quality. Default 10                                                                                                                             |
| sam_flag  | SAM FLAG to filter reads. Default 1024                                                                                                              |
| op        | Output file basename. Default parses from BAM file                                                                                                  |
| fa        | Indexed fasta file. If provided, extracts and adds reference base to the output tsv.                                                                |
| nthreads  | Number of threads to use. Each BAM file will be launched on a separate thread. Works only on Unix and macOS.                                        |

---

cancerhotspots

*Genotype known cancer hotspots from the tumor BAM file*


---

**Description**

‘cancerhotspots’ allows rapid genotyping of known somatic variants from the tumor BAM files. This facilitates to get a quick overlook of known somatic hot-spots in a matter of minutes, without spending hours on variant calling and annotation. In simple words, it fetches nucleotide frequencies of known somatic hotspots and prioritizes them based on allele frequency. Output includes a browsable/sharable HTML report of candidate variants. Known cancerhotspots for both GRCh37 and GRCh38 assemblies (3180 variants) are included. This should be sufficient and cover most of the known driver genes/events. See Reference for details.

**Usage**

```
cancerhotspots(
  bam = NULL,
  refbuild = "GRCh37",
  mapq = 10,
  sam_flag = 1024,
  vaf = 0.05,
  t_depth = 30,
  t_alt_count = 8,
  op = NULL,
  fa = NULL,
  browse = FALSE
)
```

## Arguments

|             |                                                                                      |
|-------------|--------------------------------------------------------------------------------------|
| bam         | Input bam file. Required.                                                            |
| refbuild    | Default "GRCh37". Can be "GRCh37", "GRCh38", "hg19", "hg38"                          |
| mapq        | Map quality. Default 10                                                              |
| sam_flag    | SAM FLAG to filter reads. Default 1024                                               |
| vaf         | VOF threshold. Default 0.05 [Variant filter]                                         |
| t_depth     | Depth of coverage threshold. Default 30 [Variant filter]                             |
| t_alt_count | Min. number of reads supporting tumor allele . Default 8 [Variant filter]            |
| op          | Output file basename. Default parses from BAM file                                   |
| fa          | Indexed fasta file. If provided, extracts and adds reference base to the output tsv. |
| browse      | If TRUE opens the html file in browser                                               |

## References

Chang MT, Asthana S, Gao SP, et al. Identifying recurrent mutations in cancer reveals widespread lineage diversity and mutational specificity. Nat Biotechnol. 2016;34(2):155-163. doi:10.1038/nbt.3391

## See Also

[cancerhotspotsAggr](#)

---

|                    |                                         |
|--------------------|-----------------------------------------|
| cancerhotspotsAggr | <i>Aggregate cancerhotspots reports</i> |
|--------------------|-----------------------------------------|

---

## Description

Takes tsv files generated by [cancerhotspots](#) and aggregates them into an MAF for downstream analysis

## Usage

```
cancerhotspotsAggr(  
  tsvs = NULL,  
  minVaf = 0.02,  
  minDepth = 15,  
  sampleNames = NULL,  
  maf = TRUE,  
  ...  
)
```

**Arguments**

|             |                                                                           |
|-------------|---------------------------------------------------------------------------|
| tsvs        | TSV files generated by <a href="#">cancerhotspots</a>                     |
| minVaf      | Min. VAF threshold. Default 0.02                                          |
| minDepth    | Min. depth of coverage. Default 15                                        |
| sampleNames | samples for each tsv file. Default NULL. Parses from file names.          |
| maf         | Return as an MAF object. Default TRUE.                                    |
| ...         | Additional arguments passed to <a href="#">read.maf</a> if 'maf' is TRUE. |

**Value**

[MAF](#) object

**See Also**

[cancerhotspots](#)

---

|                    |                                                                              |
|--------------------|------------------------------------------------------------------------------|
| clinicalEnrichment | <i>Performs mutational enrichment analysis for a given clinical feature.</i> |
|--------------------|------------------------------------------------------------------------------|

---

**Description**

Performs pairwise and groupwise fisher exact tests to find differentially enriched genes for every factor within a clinical feature.

**Usage**

```
clinicalEnrichment(
  maf,
  clinicalFeature = NULL,
  annotationDat = NULL,
  minMut = 5,
  useCNV = TRUE,
  pathways = FALSE
)
```

**Arguments**

|                 |                                                                                                                                                                                  |
|-----------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| maf             | <a href="#">MAF</a> object                                                                                                                                                       |
| clinicalFeature | columns names from 'clinical.data' slot of MAF to be analysed for.                                                                                                               |
| annotationDat   | If MAF file was read without clinical data, provide a custom data.frame or a tsv file with a column containing Tumor_Sample_Barcodes along with clinical features. Default NULL. |
| minMut          | Consider only genes with minimum this number of samples mutated. Default 5.                                                                                                      |
| useCNV          | whether to include copy number events if available. Default TRUE. Not applicable when 'pathways = TRUE'                                                                          |
| pathways        | Summarize genes by pathways before comparing. Default 'FALSE'                                                                                                                    |

**Details**

Performs fishers test on 2x2 contingency table for WT/Mutants in group of interest vs rest of the sample. Odds Ratio indicate the odds of observing mutant in the group of interest compared to wild-type

**Value**

result list containing p-values

**See Also**

[plotEnrichmentResults](#)

**Examples**

```
## Not run:
laml.maf = system.file('extdata', 'tcga_laml.maf.gz', package = 'maftools')
laml.clin = system.file('extdata', 'tcga_laml_annot.tsv', package = 'maftools')
laml = read.maf(maf = laml.maf, clinicalData = laml.clin)
clinicalEnrichment(laml, 'FAB_classification')

## End(Not run)
```

---

coBarplot

*Draw two barplots side by side for cohort comparison.*

---

**Description**

Draw two barplots side by side for cohort comparison.

**Usage**

```
coBarplot(
  m1,
  m2,
  genes = NULL,
  orderBy = NULL,
  m1Name = NULL,
  m2Name = NULL,
  colors = NULL,
  normalize = TRUE,
  yLims = NULL,
  borderCol = "gray",
  titleSize = 1,
  geneSize = 0.8,
  showPct = TRUE,
  pctSize = 0.7,
  axisSize = 0.8,
```

```

    showLegend = TRUE,
    legendTxtSize = 1,
    geneMar = 4
  )

```

## Arguments

|               |                                                                                               |
|---------------|-----------------------------------------------------------------------------------------------|
| m1            | first <a href="#">MAF</a> object                                                              |
| m2            | second <a href="#">MAF</a> object                                                             |
| genes         | genes to be drawn. Default takes top 5 mutated genes.                                         |
| orderBy       | Order genes by mutation rate in 'm1' or 'm2'. Default 'NULL', keeps the same order of 'genes' |
| m1Name        | optional name for first cohort                                                                |
| m2Name        | optional name for second cohort                                                               |
| colors        | named vector of colors for each Variant_Classification.                                       |
| normalize     | Default TRUE.                                                                                 |
| yLims         | Default NULL. Auto estimates. Maximum values for 'm1' and 'm2' respectively                   |
| borderCol     | Default gray                                                                                  |
| titleSize     | Default 1                                                                                     |
| geneSize      | Default 0.8                                                                                   |
| showPct       | Default TRUE                                                                                  |
| pctSize       | Default 0.7                                                                                   |
| axisSize      | Default 0.8                                                                                   |
| showLegend    | Default TRUE.                                                                                 |
| legendTxtSize | Default 0.8                                                                                   |
| geneMar       | Default 4                                                                                     |

## Details

Draws two barplots side by side to display difference between two cohorts.

## Value

Returns nothing. Just draws plot.

## Examples

```

#' ##Primary and Relapse APL
primary.apl <- system.file("extdata", "APL_primary.maf.gz", package = "maftools")
relapse.apl <- system.file("extdata", "APL_relapse.maf.gz", package = "maftools")
##Read mafs
primary.apl <- read.maf(maf = primary.apl)
relapse.apl <- read.maf(maf = relapse.apl)
##Plot
coBarplot(m1 = primary.apl, m2 = relapse.apl, m1Name = 'Primary APL', m2Name = 'Relapse APL')
dev.off()

```

---

|                   |                                                                                    |
|-------------------|------------------------------------------------------------------------------------|
| compareSignatures | <i>Compares identified denovo mutational signatures to known COSMIC signatures</i> |
|-------------------|------------------------------------------------------------------------------------|

---

### Description

Takes results from [extractSignatures](#) and compares them known COSMIC signatures. Two COSMIC databases are used for comparisons - "legacy" which includes 30 signatures, and "SBS" - which includes updated/refined 65 signatures

### Usage

```
compareSignatures(nmfRes, sig_db = "legacy", verbose = TRUE)
```

### Arguments

|         |                                                |
|---------|------------------------------------------------|
| nmfRes  | results from <a href="#">extractSignatures</a> |
| sig_db  | can be legacy or SBS. Default legacy           |
| verbose | Default TRUE                                   |

### Details

SBS signature database was obtained from <https://www.synapse.org/#!/Synapse:syn11738319.7>

### Value

list containing cosine similarities, aetiologies if available, and best match.

### See Also

[trinucleotideMatrix](#) [extractSignatures](#) [plotSignatures](#)

---

|            |                                                               |
|------------|---------------------------------------------------------------|
| coOncoplot | <i>Draw two oncoplots side by side for cohort comparison.</i> |
|------------|---------------------------------------------------------------|

---

### Description

Draw two oncoplots side by side for cohort comparison.

**Usage**

```
coOncoplot(  
  m1,  
  m2,  
  genes = NULL,  
  m1Name = NULL,  
  m2Name = NULL,  
  clinicalFeatures1 = NULL,  
  clinicalFeatures2 = NULL,  
  annotationColor1 = NULL,  
  annotationColor2 = NULL,  
  annotationFontSize = 1.2,  
  sortByAnnotation1 = FALSE,  
  annotationOrder1 = NULL,  
  sortByAnnotation2 = FALSE,  
  annotationOrder2 = NULL,  
  sampleOrder1 = NULL,  
  sampleOrder2 = NULL,  
  additionalFeature1 = NULL,  
  additionalFeaturePch1 = 20,  
  additionalFeatureCol1 = "white",  
  additionalFeatureCex1 = 0.9,  
  additionalFeature2 = NULL,  
  additionalFeaturePch2 = 20,  
  additionalFeatureCol2 = "white",  
  additionalFeatureCex2 = 0.9,  
  sepwd_genes1 = 0.5,  
  sepwd_samples1 = 0.5,  
  sepwd_genes2 = 0.5,  
  sepwd_samples2 = 0.5,  
  colors = NULL,  
  removeNonMutated = TRUE,  
  anno_height = 2,  
  legend_height = 4,  
  geneNamefont = 0.8,  
  showSampleNames = FALSE,  
  SampleNamefont = 0.5,  
  barcode_mar = 1,  
  outer_mar = 3,  
  gene_mar = 1,  
  legendFontSize = 1.2,  
  titleFontSize = 1.5,  
  keepGeneOrder = FALSE,  
  bgCol = "#CCCCCC",  
  borderCol = "white"  
)
```

**Arguments**

|                       |                                                                                                                                             |
|-----------------------|---------------------------------------------------------------------------------------------------------------------------------------------|
| m1                    | first MAF object                                                                                                                            |
| m2                    | second MAF object                                                                                                                           |
| genes                 | draw these genes. Default plots top 5 mutated genes from two cohorts.                                                                       |
| m1Name                | optional name for first cohort                                                                                                              |
| m2Name                | optional name for second cohort                                                                                                             |
| clinicalFeatures1     | columns names from 'clinical.data' slot of m1 MAF to be drawn in the plot. Default NULL.                                                    |
| clinicalFeatures2     | columns names from 'clinical.data' slot of m2 MAF to be drawn in the plot. Default NULL.                                                    |
| annotationColor1      | list of colors to use for 'clinicalFeatures1' Default NULL.                                                                                 |
| annotationColor2      | list of colors to use for 'clinicalFeatures2' Default NULL.                                                                                 |
| annotationFontSize    | font size for annotations Default 1.2                                                                                                       |
| sortByAnnotation1     | logical sort oncomatrix (samples) by provided 'clinicalFeatures1'. Sorts based on first 'clinicalFeatures1'. Defaults to FALSE. column-sort |
| annotationOrder1      | Manually specify order for annotations for 'clinicalFeatures1'. Works only for first value. Default NULL.                                   |
| sortByAnnotation2     | same as above but for m2                                                                                                                    |
| annotationOrder2      | Manually specify order for annotations for 'clinicalFeatures2'. Works only for first value. Default NULL.                                   |
| sampleOrder1          | Manually speify sample names in m1 for oncolplot ordering. Default NULL.                                                                    |
| sampleOrder2          | Manually speify sample names in m2 for oncolplot ordering. Default NULL.                                                                    |
| additionalFeature1    | a vector of length two indicating column name in the MAF and the factor level to be highlighted.                                            |
| additionalFeaturePch1 | Default 20                                                                                                                                  |
| additionalFeatureCol1 | Default "white"                                                                                                                             |
| additionalFeatureCex1 | Default 0.9                                                                                                                                 |
| additionalFeature2    | a vector of length two indicating column name in the MAF and the factor level to be highlighted.                                            |
| additionalFeaturePch2 | Default 20                                                                                                                                  |

```

additionalFeatureCol2      Default "white"
additionalFeatureCex2      Default 0.9
sepwd_genes1              Default 0.5
sepwd_samples1            Default 0.5
sepwd_genes2              Default 0.5
sepwd_samples2            Default 0.5
colors                    named vector of colors for each Variant_Classification.
removeNonMutated          Logical. If TRUE removes samples with no mutations in the oncoplot for better
                           visualization. Default TRUE.
anno_height               Height of clinical margin. Default 2
legend_height             Height of legend margin. Default 4
geneNamefont              font size for gene names. Default 1
showSampleNames           whether to show sample names. Default FALSE.
SampleNamefont            font size for sample names. Default 0.5
barcode_mar               Margin width for sample names. Default 1
outer_mar                 Margin width for outer. Default 3
gene_mar                  Margin width for gene names. Default 1
legendFontSize            font size for legend. Default 1.2
titleFontSize             font size for title. Default 1.5
keepGeneOrder             force the resulting plot to use the order of the genes as specified. Default FALSE
bgCol                     Background grid color for wild-type (not-mutated) samples. Default gray -
                           "#CCCCCC"
borderCol                 border grid color for wild-type (not-mutated) samples. Default 'white'

```

## Details

Draws two oncoplots side by side to display difference between two cohorts.

## Value

Invisibly returns a list of sample names in their order of occurrences in M1 and M2 respectively.

## Examples

```

#' ##Primary and Relapse APL
primary.apl <- system.file("extdata", "APL_primary.maf.gz", package = "maftools")
relapse.apl <- system.file("extdata", "APL_relapse.maf.gz", package = "maftools")
##Read mafs
primary.apl <- read.maf(maf = primary.apl)
relapse.apl <- read.maf(maf = relapse.apl)
##Plot
coOncoplot(m1 = primary.apl, m2 = relapse.apl, m1Name = 'Primary APL', m2Name = 'Relapse APL')
dev.off()

```

---

|                  |                               |
|------------------|-------------------------------|
| drugInteractions | <i>Drug-Gene Interactions</i> |
|------------------|-------------------------------|

---

## Description

Checks for drug-gene interactions and druggable categories

## Usage

```
drugInteractions(  
  maf,  
  top = 20,  
  genes = NULL,  
  plotType = "bar",  
  drugs = FALSE,  
  fontSize = 0.8  
)
```

## Arguments

|          |                                                                     |
|----------|---------------------------------------------------------------------|
| maf      | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a> |
| top      | Top number genes to check for. Default 20                           |
| genes    | Manually specify gene list                                          |
| plotType | Can be bar, pie Default bar plot.                                   |
| drugs    | Check for known/reported drugs. Default FALSE                       |
| fontSize | Default 0.8                                                         |

## Details

This function takes a list of genes and checks for known/reported drug-gene interactions or Drug-gable categories. All gene-drug interactions and drug claims are compiled from Drug Gene Interaction Databse. See reference for details and cite it if you use this function.

## References

Griffith, M., Griffith, O. L., Coffman, A. C., Weible, J. V., McMichael, J. F., Spies, N. C., et. al., 2013. DGIdb - Mining the druggable genome. Nature Methods.

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")  
laml <- read.maf(maf = laml.maf)  
drugInteractions(maf = laml)
```

---

|                    |                                                                             |
|--------------------|-----------------------------------------------------------------------------|
| estimateSignatures | <i>Estimate number of signatures based on cophenetic correlation metric</i> |
|--------------------|-----------------------------------------------------------------------------|

---

## Description

Estimate number of signatures based on cophenetic correlation metric

## Usage

```
estimateSignatures(
  mat,
  nMin = 2,
  nTry = 6,
  nrun = 10,
  parallel = 4,
  pConstant = NULL,
  verbose = TRUE,
  plotBestFitRes = FALSE
)
```

## Arguments

|                |                                                                                                                  |
|----------------|------------------------------------------------------------------------------------------------------------------|
| mat            | Input matrix of dimension nx96 generated by <a href="#">trinucleotideMatrix</a>                                  |
| nMin           | Minimum number of signatures to try. Default 2.                                                                  |
| nTry           | Maximum number of signatures to try. Default 6.                                                                  |
| nrun           | numeric giving the number of run to perform for each value in range. Default 5                                   |
| parallel       | Default 4. Number of cores to use.                                                                               |
| pConstant      | A small positive value to add to the matrix. Use it ONLY if the functions throws an non-conformable arrays error |
| verbose        | Default TRUE                                                                                                     |
| plotBestFitRes | plots consensus heatmap for range of values tried. Default FALSE                                                 |

## Details

This function decomposes a non-negative matrix into n signatures. Extracted signatures are compared against 30 experimentally validated signatures by calculating cosine similarity. See <http://cancer.sanger.ac.uk/cosmic/signatures> for details.

## Value

a list with NMF .rank object and summary stats.

## See Also

[plotCophenetic](#) [extractSignatures](#) [trinucleotideMatrix](#)

## Examples

```
## Not run:
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.tnm <- trinucleotideMatrix(maf = laml, ref_genome = 'BSgenome.Hsapiens.UCSC.hg19', prefix = 'chr',
add = TRUE, useSyn = TRUE)
library("NMF")
laml.sign <- estimateSignatures(mat = laml.tnm, plotBestFitRes = FALSE, nMin = 2, nTry = 3, nrun = 2, pConstant = 0.001)

## End(Not run)
```

---

|                   |                                                                  |
|-------------------|------------------------------------------------------------------|
| extractSignatures | <i>Extract mutational signatures from trinucleotide context.</i> |
|-------------------|------------------------------------------------------------------|

---

## Description

Decompose a matrix of 96 substitution classes into n signatures.

## Usage

```
extractSignatures(
  mat,
  n = NULL,
  plotBestFitRes = FALSE,
  parallel = 4,
  pConstant = NULL
)
```

## Arguments

|                |                                                                                                                                                                                  |
|----------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| mat            | Input matrix of dimension nx96 generated by <a href="#">trinucleotideMatrix</a>                                                                                                  |
| n              | decompose matrix into n signatures. Default NULL. Tries to predict best value for n by running NMF on a range of values and chooses based on cophenetic correlation coefficient. |
| plotBestFitRes | plots consensus heatmap for range of values tried. Default FALSE                                                                                                                 |
| parallel       | Default 4. Number of cores to use.                                                                                                                                               |
| pConstant      | A small positive value to add to the matrix. Use it ONLY if the functions throws an non-conformable arrays error                                                                 |

## Details

This function decomposes a non-negative matrix into n signatures.

## Value

a list with decomposed scaled signatures, signature contributions in each sample and NMF object.

**See Also**

[trinucleotideMatrix](#) [plotSignatures](#) [compareSignatures](#)

**Examples**

```
## Not run:
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.tnm <- trinucleotideMatrix(maf = laml, ref_genome = 'BSgenome.Hsapiens.UCSC.hg19', prefix = 'chr',
add = TRUE, useSyn = TRUE)
library("NMF")
laml.sign <- extractSignatures(mat = laml.tnm, plotBestFitRes = FALSE, n = 2, pConstant = 0.01)

## End(Not run)
```

---

filterMaf

---

*Filter MAF objects*


---

**Description**

Filter MAF by genes or samples

**Usage**

```
filterMaf(maf, genes = NULL, tsb = NULL, isTCGA = FALSE)
```

**Arguments**

|        |                                                     |
|--------|-----------------------------------------------------|
| maf    | an MAF object generated by <a href="#">read.maf</a> |
| genes  | remove these genes                                  |
| tsb    | remove these samples (Tumor Sample Barcodes)        |
| isTCGA | FALSE                                               |

**Value**

Filtered object of class [MAF-class](#)

**See Also**

[subsetMaf](#)

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
#get rid of samples of interest
filterMaf(maf = laml, tsb = c("TCGA-AB-2830", "TCGA-AB-2804"))
#remove genes of intrest
filterMaf(maf = laml, genes =c("TTN", "AHNAK2"))
```

---

forestPlot*Draw forest plot for differences between cohorts.*

---

## Description

Draw forest plot for differences between cohorts.

## Usage

```
forestPlot(  
  mafCompareRes,  
  pVal = 0.05,  
  fdr = NULL,  
  color = c("maroon", "royalblue"),  
  geneFontSize = 0.8,  
  titleSize = 1.2,  
  lineWidth = 1  
)
```

## Arguments

|               |                                                                       |
|---------------|-----------------------------------------------------------------------|
| mafCompareRes | results from <a href="#">mafCompare</a>                               |
| pVal          | p-value threshold. Default 0.05.                                      |
| fdr           | fdr threshold. Default NULL. If provided uses adjusted pvalues (fdr). |
| color         | vector of two colors for the lines. Default 'maroon' and 'royalblue'  |
| geneFontSize  | Font size for gene symbols. Default 0.8                               |
| titleSize     | font size for titles. Default 1.2                                     |
| lineWidth     | line width for CI bars. Default 1                                     |

## Details

Plots results from `link{mafCompare}` as a forest plot with x-axis as log10 converted odds ratio and differentially mutated genes on y-axis.

## Value

Nothing

## See Also

[mafCompare](#)

## Examples

```
##Primary and Relapse APL
primary.apl <- system.file("extdata", "APL_primary.maf.gz", package = "maftools")
relapse.apl <- system.file("extdata", "APL_relapse.maf.gz", package = "maftools")
##Read mafs
primary.apl <- read.maf(maf = primary.apl)
relapse.apl <- read.maf(maf = relapse.apl)
##Perform analysis and draw forest plot.
pt.vs.rt <- mafCompare(m1 = primary.apl, m2 = relapse.apl, m1Name = 'Primary',
m2Name = 'Relapse', minMut = 5)
forestPlot(mafCompareRes = pt.vs.rt)
```

---

|                 |                                                                          |
|-----------------|--------------------------------------------------------------------------|
| genesToBarcodes | <i>Extracts Tumor Sample Barcodes where the given genes are mutated.</i> |
|-----------------|--------------------------------------------------------------------------|

---

## Description

Extracts Tumor Sample Barcodes where the given genes are mutated.

## Usage

```
genesToBarcodes(maf, genes = NULL, justNames = FALSE, verbose = TRUE)
```

## Arguments

|           |                                                                     |
|-----------|---------------------------------------------------------------------|
| maf       | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a> |
| genes     | Hugo_Symbol for which sample names to be extracted.                 |
| justNames | if TRUE, just returns samples names instead of summarized tables.   |
| verbose   | Default TRUE                                                        |

## Value

list of `data.tables` with samples in which given genes are mutated.

## Examples

```
lam1.maf <- system.file("extdata", "tcga_lam1.maf.gz", package = "maftools")
lam1 <- read.maf(maf = lam1.maf)
genesToBarcodes(maf = lam1, genes = 'DNMT3A')
```

---

|                |                                                    |
|----------------|----------------------------------------------------|
| genotypeMatrix | <i>Creates a Genotype Matrix for every variant</i> |
|----------------|----------------------------------------------------|

---

## Description

Creates a Genotype matrix using allele frequencies or by mutation status.

## Usage

```
genotypeMatrix(  
  maf,  
  genes = NULL,  
  tsb = NULL,  
  includeSyn = FALSE,  
  vafCol = NULL,  
  vafCutoff = c(0.1, 0.75)  
)
```

## Arguments

|            |                                                                                                                                                         |
|------------|---------------------------------------------------------------------------------------------------------------------------------------------------------|
| maf        | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                     |
| genes      | create matrix for only these genes. Define NULL                                                                                                         |
| tsb        | create matrix for only these tumor sample barcodes/samples. Define NULL                                                                                 |
| includeSyn | whether to include silent mutations. Default FALSE                                                                                                      |
| vafCol     | specify column name for vaf's. Default NULL. If not provided simply assumes all mutations are heterozygous.                                             |
| vafCutoff  | specify minimum and maximum vaf to define mutations as heterozygous. Default range 0.1 to 0.75. Mutations above maximum vafs are defined as homozygous. |

## Value

matrix

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")  
laml <- read.maf(maf = laml.maf)  
genotypeMatrix(maf = laml, genes = "RUNX1")
```

---

|                 |                                            |
|-----------------|--------------------------------------------|
| getClinicalData | <i>extract annotations from MAF object</i> |
|-----------------|--------------------------------------------|

---

**Description**

extract annotations from MAF object

**Usage**

```
getClinicalData(x)

## S4 method for signature 'MAF'
getClinicalData(x)
```

**Arguments**

x                      An object of class MAF

**Value**

annotations associated with samples in MAF

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
getClinicalData(x = laml)
```

---

|                    |                                                    |
|--------------------|----------------------------------------------------|
| getCytobandSummary | <i>extract cytoband summary from GISTIC object</i> |
|--------------------|----------------------------------------------------|

---

**Description**

extract cytoband summary from GISTIC object

**Usage**

```
getCytobandSummary(x)

## S4 method for signature 'GISTIC'
getCytobandSummary(x)
```

**Arguments**

x                      An object of class GISTIC

**Value**

summarized gistic results by altered cytobands.

**Examples**

```
all.lesions <- system.file("extdata", "all_lesions.conf_99.txt", package = "maftools")
amp.genes <- system.file("extdata", "amp_genes.conf_99.txt", package = "maftools")
del.genes <- system.file("extdata", "del_genes.conf_99.txt", package = "maftools")
scores.gistic <- system.file("extdata", "scores.gistic", package = "maftools")
laml.gistic = readGistic(gisticAllLesionsFile = all.lesions, gisticAmpGenesFile = amp.genes, gisticDelGenesFile =
getCytobandSummary(laml.gistic)
```

---

getFields

---

*extract available fields from MAF object*


---

**Description**

extract available fields from MAF object

**Usage**

```
getFields(x)

## S4 method for signature 'MAF'
getFields(x)
```

**Arguments**

x                      An object of class MAF

**Value**

Field names in MAF file

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
getFields(x = laml)
```

---

|                |                                                       |
|----------------|-------------------------------------------------------|
| getGeneSummary | <i>extract gene summary from MAF or GISTIC object</i> |
|----------------|-------------------------------------------------------|

---

### Description

extract gene summary from MAF or GISTIC object

### Usage

```
getGeneSummary(x)

## S4 method for signature 'MAF'
getGeneSummary(x)

## S4 method for signature 'GISTIC'
getGeneSummary(x)
```

### Arguments

x                      An object of class MAF or GISTIC

### Value

gene summary table

### Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
getGeneSummary(laml)
```

---

|                  |                                                         |
|------------------|---------------------------------------------------------|
| getSampleSummary | <i>extract sample summary from MAF or GISTIC object</i> |
|------------------|---------------------------------------------------------|

---

### Description

extract sample summary from MAF or GISTIC object

### Usage

```
getSampleSummary(x)

## S4 method for signature 'MAF'
getSampleSummary(x)

## S4 method for signature 'GISTIC'
getSampleSummary(x)
```

**Arguments**

x                      An object of class MAF or GISTIC

**Value**

sample summary table

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
getSampleSummary(x = laml)
```

---

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

---

**Description**

S4 class for storing summarized MAF.

**Slots**

- data data.table of summarized GISTIC file.
- cnv.summary table containing alterations per sample
- cytoband.summary table containing alterations per cytoband
- gene.summary table containing alterations per gene
- cnMatrix character matrix of dimension n\*m where n is number of genes and m is number of samples
- numericMatrix numeric matrix of dimension n\*m where n is number of genes and m is number of samples
- gis.scores gistic.scores
- summary table with basic GISTIC summary stats
- classCode mapping between numeric values in numericMatrix and copy number events.

**See Also**

[getGeneSummary](#) [getSampleSummary](#) [getCytobandSummary](#)

---

|                  |                                             |
|------------------|---------------------------------------------|
| gisticBubblePlot | <i>Plot gistic results as a bubble plot</i> |
|------------------|---------------------------------------------|

---

## Description

Plots significantly altered cytobands as a function of number samples in which it is altered and number genes it contains. Size of each bubble is according to  $-\log_{10}$  transformed q values.

## Usage

```
gisticBubblePlot(
  gistic = NULL,
  color = NULL,
  markBands = NULL,
  fdrCutoff = 0.1,
  log_y = TRUE,
  txtSize = 3
)
```

## Arguments

|           |                                                                                                                     |
|-----------|---------------------------------------------------------------------------------------------------------------------|
| gistic    | an object of class GISTIC generated by readGistic                                                                   |
| color     | colors for Amp and Del events.                                                                                      |
| markBands | any cytobands to label. Can be cytoband labels, or number of top bands to highlight. Default top 5 lowest q values. |
| fdrCutoff | fdr cutoff to use. Default 0.1                                                                                      |
| log_y     | $\log_{10}$ scale y-axis (# genes affected). Default TRUE                                                           |
| txtSize   | label size for bubbles.                                                                                             |

## Value

Nothing

## Examples

```
all.lesions <- system.file("extdata", "all_lesions.conf_99.txt", package = "maftools")
amp.genes <- system.file("extdata", "amp_genes.conf_99.txt", package = "maftools")
del.genes <- system.file("extdata", "del_genes.conf_99.txt", package = "maftools")
scores.gistic <- system.file("extdata", "scores.gistic", package = "maftools")
laml.gistic = readGistic(gisticAllLesionsFile = all.lesions, gisticAmpGenesFile = amp.genes, gisticDelGenesFile = del.genes)
gisticBubblePlot(gistic = laml.gistic, markBands = "")
```

gisticChromPlot

*Plot gistic results along linearized chromosome***Description**

A genomic plot with segments highlighting significant Amplifications and Deletion regions.

**Usage**

```
gisticChromPlot(
  gistic = NULL,
  fdrCutoff = 0.1,
  markBands = NULL,
  color = NULL,
  ref.build = "hg19",
  cytobandOffset = 0.01,
  txtSize = 0.8,
  cytobandTxtSize = 0.6,
  maf = NULL,
  mutGenes = NULL,
  y_lims = NULL,
  mutGenesTxtSize = 0.6
)
```

**Arguments**

|                 |                                                                  |
|-----------------|------------------------------------------------------------------|
| gistic          | an object of class GISTIC generated by readGistic                |
| fdrCutoff       | fdr cutoff to use. Default 0.1                                   |
| markBands       | any cytobands to label. Default top 5 lowest q values.           |
| color           | colors for Amp and Del events.                                   |
| ref.build       | reference build. Could be hg18, hg19 or hg38.                    |
| cytobandOffset  | if scores.gistic file is given use this to adjust cytoband size. |
| txtSize         | label size for labels                                            |
| cytobandTxtSize | label size for cytoband                                          |
| maf             | an optional maf object                                           |
| mutGenes        | mutated genes from maf object to be highlighted                  |
| y_lims          | Default NULL. A vector upper and lower y-axis limits             |
| mutGenesTxtSize | Default 0.6                                                      |

**Value**

nothing

**Examples**

```
all.lesions <- system.file("extdata", "all_lesions.conf_99.txt", package = "maftools")
amp.genes <- system.file("extdata", "amp_genes.conf_99.txt", package = "maftools")
del.genes <- system.file("extdata", "del_genes.conf_99.txt", package = "maftools")
scores.gistic <- system.file("extdata", "scores.gistic", package = "maftools")
laml.gistic = readGistic(gisticAllLesionsFile = all.lesions, gisticAmpGenesFile = amp.genes, gisticDelGenesFile =
gisticChromPlot(laml.gistic)
```

gisticOncoPlot

*Plot gistic results.***Description**

takes output generated by readGistic and draws a plot similar to oncoplot.

**Usage**

```
gisticOncoPlot(
  gistic = NULL,
  top = NULL,
  bands = NULL,
  showTumorSampleBarcodes = FALSE,
  gene_mar = 5,
  barcode_mar = 6,
  sepwd_genes = 0.5,
  sepwd_samples = 0.25,
  clinicalData = NULL,
  clinicalFeatures = NULL,
  sortByAnnotation = FALSE,
  sampleOrder = NULL,
  annotationColor = NULL,
  bandsToIgnore = NULL,
  removeNonAltered = TRUE,
  colors = NULL,
  SampleNameFontSize = 0.6,
  fontSize = 0.8,
  legendFontSize = 1.2,
  annotationFontSize = 1.2,
  borderCol = "white",
  bgCol = "#CCCCCC"
)
```

**Arguments**

|        |                                                                          |
|--------|--------------------------------------------------------------------------|
| gistic | an <a href="#">GISTIC</a> object generated by <a href="#">readGistic</a> |
| top    | how many top cytobands to be drawn. defaults to all.                     |
| bands  | draw oncoplot for these bands. Default NULL.                             |

|                         |                                                                                                             |
|-------------------------|-------------------------------------------------------------------------------------------------------------|
| showTumorSampleBarcodes | logical to include sample names.                                                                            |
| gene_mar                | Default 5                                                                                                   |
| barcode_mar             | Default 6                                                                                                   |
| sepwd_genes             | Default 0.5                                                                                                 |
| sepwd_samples           | Default 0.25                                                                                                |
| clinicalData            | data.frame with columns containing Tumor_Sample_Barcodes and rest of columns with annotations.              |
| clinicalFeatures        | columns names from 'clinicalData' to be drawn in the plot. Dafault NULL.                                    |
| sortByAnnotation        | logical sort oncomatrix (samples) by provided 'clinicalFeatures'. Defaults to FALSE. column-sort            |
| sampleOrder             | Manually speify sample names for oncolplot ordering. Default NULL.                                          |
| annotationColor         | list of colors to use for clinicalFeatures. Default NULL.                                                   |
| bandsToIgnore           | do not show these bands in the plot Default NULL.                                                           |
| removeNonAltered        | Logical. If TRUE removes samples with no mutations in the oncoplot for better visualization. Default FALSE. |
| colors                  | named vector of colors Amp and Del events.                                                                  |
| SampleNamefontSize      | font size for sample names. Default 0.6                                                                     |
| fontSize                | font size for cytoband names. Default 0.8                                                                   |
| legendFontSize          | font size for legend. Default 1.2                                                                           |
| annotationFontSize      | font size for annotations. Default 1.2                                                                      |
| borderCol               | Default "white"                                                                                             |
| bgCol                   | Default "#CCCCCC"                                                                                           |

## Details

Takes gistic file as input and plots it as a matrix. Any desired annotations can be added at the bottom of the oncoplot by providing annotation

## Value

None.

## See Also

[oncostrip](#)

## Examples

```
all.lesions <- system.file("extdata", "all_lesions.conf_99.txt", package = "maftools")
amp.genes <- system.file("extdata", "amp_genes.conf_99.txt", package = "maftools")
del.genes <- system.file("extdata", "del_genes.conf_99.txt", package = "maftools")
scores.gistic <- system.file("extdata", "scores.gistic", package = "maftools")
laml.gistic = readGistic(gisticAllLesionsFile = all.lesions, gisticAmpGenesFile = amp.genes, gisticDelGenesFile =
gisticOncoPlot(laml.gistic)
```

---

gtMarkers

---

*Extract read counts from genetic markers for ASCAT analysis*


---

## Description

The function will generate tsv files ‘<tumor/normal>\_nucleotide\_counts.tsv’ that can be used for downstream analysis. Note that the function will process ~900K loci from Affymetrix Genome-Wide Human SNP 6.0 Array. The process can be sped up by increasing ‘nthreads’ which will launch each chromosome on a separate thread. Currently hg19 and hg38 are supported. Files need to be further processed with [prepAscat](#) for tumor-normal pair, or [prepAscat\\_t](#) for tumor only samples.

## Usage

```
gtMarkers(
  t_bam = NULL,
  n_bam = NULL,
  build = "hg19",
  prefix = NULL,
  add = TRUE,
  mapq = 10,
  sam_flag = 1024,
  loci = NULL,
  fa = NULL,
  op = NULL,
  zerobased = FALSE,
  nthreads = 4,
  verbose = TRUE
)
```

## Arguments

|        |                                                                                                                                                                                               |
|--------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| t_bam  | Tumor BAM file. Required                                                                                                                                                                      |
| n_bam  | Normal BAM file. Recommended                                                                                                                                                                  |
| build  | Default hg19. Mutually exclusive with ‘loci’. Currently supported ‘hg19’ and ‘hg38’ and includes ca. 900K SNPs from Affymetrix Genome-Wide Human SNP 6.0 Array. SNP file has no ‘chr’ prefix. |
| prefix | Prefix to add or remove from contig names in loci file. For example, in case BAM files have ‘chr’ prefix, set prefix = ‘chr’                                                                  |

|           |                                                                                                                              |
|-----------|------------------------------------------------------------------------------------------------------------------------------|
| add       | If prefix is used, default is to add prefix to contig names in loci file. If false prefix will be removed from contig names. |
| mapq      | Minimum mapping quality. Default 10                                                                                          |
| sam_flag  | SAM FLAG to filter reads. Default 1024                                                                                       |
| loci      | A tab separated file with chr and position. If not available use 'build' argument.                                           |
| fa        | Indexed fasta file. If provided, extracts and adds reference base to the output tsv.                                         |
| op        | Output file basename. Default parses from BAM file                                                                           |
| zerobased | are coordinates zero-based. Default FALSE. Use only if 'loci' is used.                                                       |
| nthreads  | Number of threads to use. Default 4. Each chromosome will be launched on a separate thread. Works only on Unix and macOS.    |
| verbose   | Default TRUE                                                                                                                 |

**See Also**

[prepAscat](#) [prepAscat\\_t](#) [segmentLogR](#)

---

icgcSimpleMutationToMAF

*Converts ICGC Simple Somatic Mutation format file to MAF*

---

**Description**

Converts ICGC Simple Somatic Mutation format file to Mutation Annotation Format. Basic fields are converted as per MAF specifications, rest of the fields are retained as in the input file. Ensemble gene IDs are converted to HGNC Symbols. Note that by default Simple Somatic Mutation format contains all affected transcripts of a variant resulting in multiple entries of the same variant in same sample. It is hard to choose a single affected transcript based on annotations alone and by default this program removes repeated variants as duplicated entries. If you wish to keep all of them, set `removeDuplicatedVariants` to FALSE.

**Usage**

```
icgcSimpleMutationToMAF(
  icgc,
  basename = NA,
  MAFobj = FALSE,
  clinicalData = NULL,
  removeDuplicatedVariants = TRUE,
  addHugoSymbol = FALSE
)
```

**Arguments**

|                          |                                                                                                                               |
|--------------------------|-------------------------------------------------------------------------------------------------------------------------------|
| icgc                     | Input data in ICGC Simple Somatic Mutation format. Can be gz compressed.                                                      |
| basename                 | If given writes to output file with basename.                                                                                 |
| MAFobj                   | If TRUE returns results as an <a href="#">MAF</a> object.                                                                     |
| clinicalData             | Clinical data associated with each sample/Tumor_Sample_Barcode in MAF. Could be a text file or a data.frame. Default NULL.    |
| removeDuplicatedVariants | removes repeated variants in a particular sample, mapped to multiple transcripts of same Gene. See Description. Default TRUE. |
| addHugoSymbol            | If TRUE replaces ensemble gene IDs with Hugo_Symbols. Default FALSE.                                                          |

**Details**

ICGC Simple Somatic Mutation format specification can be found here: <http://docs.icgc.org/submission/guide/icgc-simple-somatic-mutation-format/>

**Value**

tab delimited MAF file.

**Examples**

```
esca.icgc <- system.file("extdata", "simple_somatic_mutation.open.ESCA-CN.sample.tsv.gz", package = "maftools")
esca.maf <- icgcSimpleMutationToMAF(icgc = esca.icgc)
```

---

|                    |                                                                     |
|--------------------|---------------------------------------------------------------------|
| inferHeterogeneity | <i>Clusters variants based on Variant Allele Frequencies (VAF).</i> |
|--------------------|---------------------------------------------------------------------|

---

**Description**

takes output generated by read.maf and clusters variants to infer tumor heterogeneity. This function requires VAF for clustering and density estimation. VAF can be on the scale 0-1 or 0-100. Optionally if copy number information is available, it can be provided as a segmented file (e.g, from Circular Binary Segmentation). Those variants in copy number altered regions will be ignored.

**Usage**

```
inferHeterogeneity(
  maf,
  tsb = NULL,
  top = 5,
  vafCol = NULL,
  segFile = NULL,
  ignChr = NULL,
  minVaf = 0,
  maxVaf = 1,
```

```

    useSyn = FALSE,
    dirichlet = FALSE
  )

```

### Arguments

|           |                                                                                                                                             |
|-----------|---------------------------------------------------------------------------------------------------------------------------------------------|
| maf       | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                         |
| tsb       | specify sample names (Tumor_Sample_Barcodes) for which clustering has to be done.                                                           |
| top       | if tsb is NULL, uses top n number of most mutated samples. Defaults to 5.                                                                   |
| vafCol    | manually specify column name for vafs. Default looks for column 't_vaf'                                                                     |
| segFile   | path to CBS segmented copy number file. Column names should be Sample, Chromosome, Start, End, Num_Probes and Segment_Mean (log2 scale).    |
| ignChr    | ignore these chromosomes from analysis. e.g, sex chromosomes chrX, chrY. Default NULL.                                                      |
| minVaf    | filter low frequency variants. Low vaf variants maybe due to sequencing error. Default 0. (on the scale of 0 to 1)                          |
| maxVaf    | filter high frequency variants. High vaf variants maybe due to copy number alterations or impure tumor. Default 1. (on the scale of 0 to 1) |
| useSyn    | Use synonymous variants. Default FALSE.                                                                                                     |
| dirichlet | Deprecated! No longer supported. uses nonparametric dirichlet process for clustering. Default FALSE - uses finite mixture models.           |

### Details

This function clusters variants based on VAF to estimate univariate density and cluster classification. There are two methods available for clustering. Default using parametric finite mixture models and another method using nonparametric infinite mixture models (Dirichlet process).

### Value

list of clustering tables.

### References

- Chris Fraley and Adrian E. Raftery (2002) Model-based Clustering, Discriminant Analysis and Density Estimation Journal of the American Statistical Association 97:611-631
- Jara A, Hanson TE, Quintana FA, Muller P, Rosner GL. DPpackage: Bayesian Semi- and Nonparametric Modeling in R. Journal of statistical software. 2011;40(5):1-30.
- Olshen AB, Venkatraman ES, Lucito R, Wigler M. Circular binary segmentation for the analysis of array-based DNA copy number data. Biostatistics. 2004;5(4):557-72.

### See Also

[plotClusters](#)

**Examples**

```
## Not run:
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
TCGA.AB.2972.clust <- inferHeterogeneity(maf = laml, tsb = 'TCGA-AB-2972', vafCol = 'i_TumorVAF_WU')

## End(Not run)
```

---

lollipopPlot

---

*Draws lollipop plot of amino acid changes on to Protein structure.*


---

**Description**

Draws lollipop plot of amino acid changes. Protein domains are derived from PFAM database.

**Usage**

```
lollipopPlot(
  maf,
  gene = NULL,
  AACol = NULL,
  labelPos = NULL,
  labPosSize = 0.9,
  showMutationRate = TRUE,
  showDomainLabel = TRUE,
  cBioPortal = FALSE,
  refSeqID = NULL,
  proteinID = NULL,
  roundedRect = TRUE,
  repel = FALSE,
  collapsePosLabel = TRUE,
  showLegend = TRUE,
  legendTxtSize = 0.8,
  labPosAngle = 0,
  domainLabelSize = 0.8,
  axisTextSize = c(1, 1),
  printCount = FALSE,
  colors = NULL,
  domainAlpha = 1,
  domainBorderCol = "black",
  bgBorderCol = "black",
  labelOnlyUniqueDoamins = TRUE,
  defaultYaxis = FALSE,
  titleSize = c(1.2, 1),
  pointSize = 1.5
)
```

**Arguments**

|                        |                                                                                                                                                                                                                                                                  |
|------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| maf                    | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                                                                                                                              |
| gene                   | HGNC symbol for which protein structure to be drawn.                                                                                                                                                                                                             |
| AACol                  | manually specify column name for amino acid changes. Default looks for fields 'HGVSp_Short', 'AACChange' or 'Protein_Change'. Changes can be of any format i.e, can be a numeric value or HGVSp annotations (e.g; p.P459L, p.L2195Pfs*30 or p.Leu2195ProfsTer30) |
| labelPos               | Amino acid positions to label. If 'all', labels all variants.                                                                                                                                                                                                    |
| labPosSize             | Text size for labels. Default 0.9                                                                                                                                                                                                                                |
| showMutationRate       | Whether to show the somatic mutation rate on the title. Default TRUE                                                                                                                                                                                             |
| showDomainLabel        | Label domains within the plot. Default TRUE. If 'FALSE' domains are annotated in legend.                                                                                                                                                                         |
| cBioPortal             | Adds annotations similar to cBioPortals MutationMapper and collapse Variants into Truncating and rest.                                                                                                                                                           |
| refSeqID               | RefSeq transcript identifier for gene if known.                                                                                                                                                                                                                  |
| proteinID              | RefSeq protein identifier for gene if known.                                                                                                                                                                                                                     |
| roundedRect            | Default TRUE. If 'TRUE' domains are drawn with rounded corners. Requires <a href="#">berryFunctions</a>                                                                                                                                                          |
| repel                  | If points are too close to each other, use this option to repel them. Default FALSE. Warning: naive method, might make plot ugly in case of too many variants!                                                                                                   |
| collapsePosLabel       | Collapses overlapping labels at same position. Default TRUE                                                                                                                                                                                                      |
| showLegend             | Default TRUE                                                                                                                                                                                                                                                     |
| legendTxtSize          | Text size for legend. Default 0.8                                                                                                                                                                                                                                |
| labPosAngle            | angle for labels. Defaults to horizontal 0 degree labels. Set to 90 for vertical; 45 for diagonal labels.                                                                                                                                                        |
| domainLabelSize        | text size for domain labels. Default 0.8                                                                                                                                                                                                                         |
| axisTextSize           | text size x and y tick labels. Default c(1,1).                                                                                                                                                                                                                   |
| printCount             | If TRUE, prints number of summarized variants for the given protein.                                                                                                                                                                                             |
| colors                 | named vector of colors for each Variant_Classification. Default NULL.                                                                                                                                                                                            |
| domainAlpha            | Default 1                                                                                                                                                                                                                                                        |
| domainBorderCol        | Default "black". Set to NA to remove.                                                                                                                                                                                                                            |
| bgBorderCol            | Default "black". Set to NA to remove.                                                                                                                                                                                                                            |
| labelOnlyUniqueDoamins | Default TRUE only labels unique doamins.                                                                                                                                                                                                                         |
| defaultYaxis           | If FALSE, just labels min and maximum y values on y axis.                                                                                                                                                                                                        |
| titleSize              | font size for title and subtitle. Default c(1.2, 1)                                                                                                                                                                                                              |
| pointSize              | size of lollipop heads. Default 1.5                                                                                                                                                                                                                              |

**Details**

This function by default looks for fields 'HGVSp\_Short', 'AACChange' or 'Protein\_Change' in maf file. One can also manually specify field name containing amino acid changes.

**Value**

Nothing

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
lollipopPlot(maf = laml, gene = 'KIT', AACol = 'Protein_Change')
```

---

lollipopPlot2

---

*Compare two lollipop plots*


---

**Description**

Compare two lollipop plots

**Usage**

```
lollipopPlot2(
  m1,
  m2,
  gene = NULL,
  AACol1 = NULL,
  AACol2 = NULL,
  m1_name = NULL,
  m2_name = NULL,
  m1_label = NULL,
  m2_label = NULL,
  refSeqID = NULL,
  proteinID = NULL,
  labPosAngle = 0,
  labPosSize = 0.9,
  colors = NULL,
  alpha = 1,
  axisTextSize = c(1, 1),
  pointSize = 1.2,
  roundedRect = TRUE,
  showDomainLabel = TRUE,
  domainBorderCol = "black",
  domainLabelSize = 1,
  legendTxtSize = 1,
  verbose = TRUE
)
```

**Arguments**

|                 |                                                                                                                                     |
|-----------------|-------------------------------------------------------------------------------------------------------------------------------------|
| m1              | first <a href="#">MAF</a> object                                                                                                    |
| m2              | second <a href="#">MAF</a> object                                                                                                   |
| gene            | HGNC symbol for which protein structure to be drawn.                                                                                |
| AACol1          | manually specify column name for amino acid changes in m1. Default looks for fields 'HGVSp_Short', 'AACChange' or 'Protein_Change'. |
| AACol2          | manually specify column name for amino acid changes in m2. Default looks for fields 'HGVSp_Short', 'AACChange' or 'Protein_Change'. |
| m1_name         | name for m1 cohort. optional.                                                                                                       |
| m2_name         | name for m2 cohort. optional.                                                                                                       |
| m1_label        | Amino acid positions to label for m1 cohort. If 'all', labels all variants.                                                         |
| m2_label        | Amino acid positions to label for m2 cohort. If 'all', labels all variants.                                                         |
| refSeqID        | RefSeq transcript identifier for gene if known.                                                                                     |
| proteinID       | RefSeq protein identifier for gene if known.                                                                                        |
| labPosAngle     | angle for labels. Defaults to horizontal 0 degree labels. Set to 90 for vertical; 45 for diagonal labels.                           |
| labPosSize      | Text size for labels. Default 3                                                                                                     |
| colors          | named vector of colors for each Variant_Classification. Default NULL.                                                               |
| alpha           | color adjustment. Default 1                                                                                                         |
| axisTextSize    | text size for axis labels. Default 1.                                                                                               |
| pointSize       | size of lollipop heads. Default 1.2                                                                                                 |
| roundedRect     | Default FALSE. If 'TRUE' domains are drawn with rounded corners. Requires <a href="#">berryFunctions</a>                            |
| showDomainLabel | Label domains within the plot. Default TRUE. If FALSE domains are annotated in legend.                                              |
| domainBorderCol | Default "black". Set to NA to remove.                                                                                               |
| domainLabelSize | text size for domain labels. Default 1.                                                                                             |
| legendTxtSize   | Default 1.                                                                                                                          |
| verbose         | Default TRUE                                                                                                                        |

**Details**

Draws lollipop plot for a gene from two cohorts

**Value**

invisible list of domain overlaps

See Also

[lollipopPlot](#)  
[mafCompare](#)

Examples

```
primary.apl <- system.file("extdata", "APL_primary.maf.gz", package = "maftools")
relapse.apl <- system.file("extdata", "APL_relapse.maf.gz", package = "maftools")
primary.apl <- read.maf(maf = primary.apl)
relapse.apl <- read.maf(maf = relapse.apl)
lollipopPlot2(m1 = primary.apl, m2 = relapse.apl, gene = "FLT3", AACol1 = "amino_acid_change", AACol2 = "amino_acid")
```

---

|     |                                |
|-----|--------------------------------|
| MAF | <i>Construct an MAF object</i> |
|-----|--------------------------------|

---

Description

Constructor function which takes non-synonymous, and synonymous variants along with an optional clinical information and generates an MAF object

Usage

```
MAF(nonSyn = NULL, syn = NULL, clinicalData = NULL, verbose = TRUE)
```

Arguments

|              |                                                                                                                                                                                            |
|--------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| nonSyn       | non-synonymous variants as a data.table or any object that can be coerced into a data.table (e.g: data.frame, GRanges)                                                                     |
| syn          | synonymous variants as a data.table or any object that can be coerced into a data.table (e.g: data.frame, GRanges)                                                                         |
| clinicalData | Clinical data associated with each sample/Tumor_Sample_Barcode in MAF. Could be a text file or a data.frame. Requires at least a column with the name ‘Tumor_Sample_Barcode‘ Default NULL. |
| verbose      | Default TRUE                                                                                                                                                                               |

Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml_dt = data.table::fread(input = laml.maf)
laml.clin = system.file('extdata', 'tcga_laml_annot.tsv', package = 'maftools') #Clinical data
# Just for demonstration
nsyn_vars = laml_dt[Variant_Classification %in% "Missense_Mutation"]
syn_vars = laml_dt[Variant_Classification %in% "Silent"]
maftools::MAF(nonSyn = nsyn_vars, syn = syn_vars, clinicalData = laml.clin)
```

---

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

---

**Description**

S4 class for storing summarized MAF.

**Slots**

- data data.table of MAF file containing all non-synonymous variants.
- variants.per.sample table containing variants per sample
- variant.type.summary table containing variant types per sample
- variant.classification.summary table containing variant classification per sample
- gene.summary table containing variant classification per gene
- summary table with basic MAF summary stats
- maf.silent subset of main MAF containing only silent variants
- clinical.data clinical data associated with each sample/Tumor\_Sample\_Barcode in MAF.

**See Also**

[getGeneSummary](#) [getSampleSummary](#) [getFields](#)

---

|         |                                                   |
|---------|---------------------------------------------------|
| maf2mae | <i>Convert MAF to MultiAssayExperiment object</i> |
|---------|---------------------------------------------------|

---

**Description**

Generates an object of class MultiAssayExperiment from MAF object

**Usage**

```
maf2mae(m = NULL)
```

**Arguments**

m                      an MAF object

**Examples**

```
laml.maf = system.file('extdata', 'tcga_laml.maf.gz', package = 'maftools')
laml.clin = system.file('extdata', 'tcga_laml_annot.tsv', package = 'maftools')
laml = read.maf(maf = laml.maf, clinicalData = laml.clin)
maf2mae(laml)
```

---

`mafbarplot`*Creates a bar plot*

---

### Description

Takes an MAF object and generates a barplot of mutated genes color coded for variant classification

### Usage

```
mafbarplot(  
  maf,  
  n = 20,  
  genes = NULL,  
  color = NULL,  
  fontSize = 0.7,  
  includeCN = FALSE,  
  legendfontSize = 0.7,  
  borderCol = "#34495e",  
  showPct = TRUE  
)
```

### Arguments

|                             |                                                                       |
|-----------------------------|-----------------------------------------------------------------------|
| <code>maf</code>            | an <a href="#">MAF</a> object                                         |
| <code>n</code>              | Number of genes to include. Default 20.                               |
| <code>genes</code>          | Manually provide names of genes. Default NULL.                        |
| <code>color</code>          | named vector of colors for each Variant_Classification. Default NULL. |
| <code>fontSize</code>       | Default 0.7                                                           |
| <code>includeCN</code>      | Include copy number events if available? Default FALSE                |
| <code>legendfontSize</code> | Default 0.7                                                           |
| <code>borderCol</code>      | Default "#34495e". Set to 'NA' for no border color.                   |
| <code>showPct</code>        | Default TRUE. Show percent altered samples.                           |

### Examples

```
laml.maf = system.file("extdata", "tcga_laml.maf.gz", package = "maftools") #MAF file  
laml = read.maf(maf = laml.maf)  
mafbarplot(maf = laml)
```

---

|            |                                   |
|------------|-----------------------------------|
| mafCompare | <i>compare two cohorts (MAF).</i> |
|------------|-----------------------------------|

---

## Description

compare two cohorts (MAF).

## Usage

```
mafCompare(
  m1,
  m2,
  m1Name = NULL,
  m2Name = NULL,
  minMut = 5,
  useCNV = TRUE,
  pathways = FALSE,
  custom_pw = NULL,
  pseudoCount = FALSE
)
```

## Arguments

|             |                                                                                                                                                                                            |
|-------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| m1          | first <a href="#">MAF</a> object                                                                                                                                                           |
| m2          | second <a href="#">MAF</a> object                                                                                                                                                          |
| m1Name      | optional name for first cohort                                                                                                                                                             |
| m2Name      | optional name for second cohort                                                                                                                                                            |
| minMut      | Consider only genes with minimum this number of samples mutated in at least one of the cohort for analysis. Helpful to ignore single mutated genes. Default 5.                             |
| useCNV      | whether to include copy number events. Default TRUE if available.. Not applicable when 'pathways = TRUE'                                                                                   |
| pathways    | Summarize genes by pathways before comparing. Default 'FALSE'                                                                                                                              |
| custom_pw   | Optional. Can be a two column data.frame/tsv-file with pathway-name and genes involved in them. Default 'NULL', uses a predefined list of pathways. Applicable only when 'pathways = TRUE' |
| pseudoCount | If TRUE, adds 1 to the contingency table with 0's to avoid 'Inf' values in the estimated odds-ratio.                                                                                       |

## Details

Performs fisher test on 2x2 contingency table generated from two cohorts to find differentially mutated genes.

## Value

result list

**See Also**[forestPlot](#)[lollipopPlot2](#)**Examples**

```

primary.apl <- system.file("extdata", "APL_primary.maf.gz", package = "maftools")
relapse.apl <- system.file("extdata", "APL_relapse.maf.gz", package = "maftools")
primary.apl <- read.maf(maf = primary.apl)
relapse.apl <- read.maf(maf = relapse.apl)
pt.vs.rt <- mafCompare(m1 = primary.apl, m2 = relapse.apl, m1Name = 'Primary',
m2Name = 'Relapse', minMut = 5)

```

mafSummary

*Summary statistics of MAF***Description**

Summarizes genes and samples irrespective of the type of alteration. This is different from [getSampleSummary](#) and [getGeneSummary](#) which returns summaries of only non-synonymous variants.

**Usage**

```
mafSummary(maf)
```

**Arguments**

maf                    an MAF object generated by [read.maf](#)

**Details**

This function takes MAF object as input and returns summary table.

**Value**

Returns a list of summarized tables

**See Also**

[getGeneSummary](#) [getSampleSummary](#)

**Examples**

```

laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
mafSummary(maf = laml)

```

---

|              |                                                 |
|--------------|-------------------------------------------------|
| mafSurvGroup | <i>Performs survival analysis for a geneset</i> |
|--------------|-------------------------------------------------|

---

## Description

Similar to [mafSurvival](#) but for a geneset

## Usage

```
mafSurvGroup(  
  maf,  
  geneSet = NULL,  
  minMut = NA,  
  clinicalData = NULL,  
  time = "Time",  
  Status = "Status"  
)
```

## Arguments

|              |                                                                                                                                                                   |
|--------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| maf          | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                               |
| geneSet      | gene names for which survival analysis needs to be performed.                                                                                                     |
| minMut       | minimum number of mutated genes in the 'geneSet' to consider a sample as a mutant. Default, 'NA', samples with all the genes mutated are treated as mutant group. |
| clinicalData | dataframe containing events and time to events. Default looks for clinical data in annotation slot of <a href="#">MAF</a> .                                       |
| time         | column name containing time in clinicalData                                                                                                                       |
| Status       | column name containing status of patients in clinicalData. must be logical or numeric. e.g, TRUE or FALSE, 1 or 0.                                                |

## Value

Survival plot

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")  
laml.clin <- system.file("extdata", "tcga_laml_annot.tsv", package = "maftools")  
laml <- read.maf(maf = laml.maf, clinicalData = laml.clin)  
mafSurvGroup(maf = laml, geneSet = c('DNMT3A', 'FLT3'), time = 'days_to_last_followup', Status = 'Overall_Survival')
```

mafSurvival

*Performs survival analysis***Description**

Performs survival analysis by grouping samples from maf based on mutation status of given gene(s) or manual grouping of samples.

**Usage**

```
mafSurvival(
  maf,
  genes = NULL,
  samples = NULL,
  clinicalData = NULL,
  time = "Time",
  Status = "Status",
  groupNames = c("Mutant", "WT"),
  showConfInt = TRUE,
  addInfo = TRUE,
  col = c("maroon", "royalblue"),
  isTCGA = FALSE,
  textSize = 12
)
```

**Arguments**

|              |                                                                                                                                            |
|--------------|--------------------------------------------------------------------------------------------------------------------------------------------|
| maf          | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                        |
| genes        | gene names for which survival analysis needs to be performed. Samples with mutations in any one of the genes provided are used as mutants. |
| samples      | samples to group by. Genes and samples are mutually exclusive.                                                                             |
| clinicalData | dataframe containing events and time to events. Default looks for clinical data in annotation slot of <a href="#">MAF</a> .                |
| time         | column name containing time in clinicalData                                                                                                |
| Status       | column name containing status of patients in clinicalData. must be logical or numeric. e.g, TRUE or FALSE, 1 or 0.                         |
| groupNames   | names for groups. Should be of length two. Default c("Mutant", "WT")                                                                       |
| showConfInt  | TRUE. Whether to show confidence interval in KM plot.                                                                                      |
| addInfo      | TRUE. Whether to show survival info in the plot.                                                                                           |
| col          | colors for plotting.                                                                                                                       |
| isTCGA       | FALSE. Is data is from TCGA.                                                                                                               |
| textSize     | Text size for surv table. Default 7.                                                                                                       |

## Details

This function takes MAF file and groups them based on mutation status associated with given gene(s) and performs survival analysis. Requires dataframe containing survival status and time to event. Make sure sample names match to Tumor Sample Barcodes from MAF file.

## Value

Survival plot

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml.clin <- system.file("extdata", "tcga_laml_annot.tsv", package = "maftools")
laml <- read.maf(maf = laml.maf, clinicalData = laml.clin)
mafSurvival(maf = laml, genes = 'DNMT3A', time = 'days_to_last_followup', Status = 'Overall_Survival_Status', isTCGA = TRUE)
```

---

|            |                                                                   |
|------------|-------------------------------------------------------------------|
| math.score | <i>calculates MATH (Mutant-Allele Tumor Heterogeneity) score.</i> |
|------------|-------------------------------------------------------------------|

---

## Description

calculates MATH scores from variant allele frequencies. Mutant-Allele Tumor Heterogeneity (MATH) score is a measure of intra-tumor genetic heterogeneity. High MATH scores are related to lower survival rates. This function requires vafs.

## Usage

```
math.score(maf, vafCol = NULL, sampleName = NULL, vafCutoff = 0.075)
```

## Arguments

|            |                                                                                         |
|------------|-----------------------------------------------------------------------------------------|
| maf        | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                     |
| vafCol     | manually specify column name for vafs. Default looks for column 't_vaf'                 |
| sampleName | sample name for which MATH score to be calculated. If NULL, calculates for all samples. |
| vafCutoff  | minimum vaf for a variant to be considered for score calculation. Default 0.075         |

## Value

data.table with MATH score for every Tumor\_Sample\_Barcode

## References

Mroz, Edmund A. et al. Intra-Tumor Genetic Heterogeneity and Mortality in Head and Neck Cancer: Analysis of Data from The Cancer Genome Atlas. Ed. Andrew H. Beck. PLoS Medicine 12.2 (2015): e1001786.

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.math <- math.score(maf = laml, vafCol = 'i_TumorVAF_WU',
  sampleName = c('TCGA-AB-3009', 'TCGA-AB-2849', 'TCGA-AB-3002', 'TCGA-AB-2972'))
```

---

merge\_mafs

*Merge multiple mafs into single MAF*


---

**Description**

Merges multiple maf files/objects/data.frames into a single MAF.

**Usage**

```
merge_mafs(mafs, verbose = TRUE, ...)
```

**Arguments**

|         |                                                                             |
|---------|-----------------------------------------------------------------------------|
| mafs    | a list of <a href="#">MAF</a> objects or data.frames or paths to MAF files. |
| verbose | Default TRUE                                                                |
| ...     | additional arguments passed <a href="#">read.maf</a>                        |

**Value**

[MAF](#) object

---

mutCountMatrix

*Generates count matrix of mutations.*


---

**Description**

Generates a count matrix of mutations. i.e, number of mutations per gene per sample.

**Usage**

```
mutCountMatrix(
  maf,
  includeSyn = FALSE,
  countOnly = NULL,
  removeNonMutated = TRUE
)
```

**Arguments**

|                               |                                                                                                                                                                                                    |
|-------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>maf</code>              | an MAF object generated by <a href="#">read.maf</a>                                                                                                                                                |
| <code>includeSyn</code>       | whether to include synonymous variants in output matrix. Default FALSE                                                                                                                             |
| <code>countOnly</code>        | Default NULL - counts all variants. You can specify type of 'Variant_Classification' to count. For e.g, <code>countOnly = 'Splice_Site'</code> will generate matrix for only Splice_Site variants. |
| <code>removeNonMutated</code> | Logical Default TRUE, removes samples with no mutations from the matrix.                                                                                                                           |

**Value**

Integer Matrix

**See Also**

[getFields](#) [getGeneSummary](#) [getSampleSummary](#)

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
##Generate matrix
mutCountMatrix(maf = laml)
##Generate count matrix of Splice_Site mutations
mutCountMatrix(maf = laml, countOnly = 'Splice_Site')
```

---

oncodrive

---

*Detect cancer driver genes based on positional clustering of variants.*


---

**Description**

Clusters variants based on their position to detect disease causing genes.

**Usage**

```
oncodrive(
  maf,
  AACol = NULL,
  minMut = 5,
  pvalMethod = "zscore",
  nBgGenes = 100,
  bgEstimate = TRUE,
  ignoreGenes = NULL
)
```

## Arguments

|                          |                                                                                                                                                               |
|--------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>maf</code>         | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                           |
| <code>AACol</code>       | manually specify column name for amino acid changes. Default looks for field 'AACChange'                                                                      |
| <code>minMut</code>      | minimum number of mutations required for a gene to be included in analysis. Default 5.                                                                        |
| <code>pvalMethod</code>  | either zscore (default method for oncodriveCLUST), poisson or combined (uses lowest of the two pvalues).                                                      |
| <code>nBgGenes</code>    | minimum number of genes required to estimate background score. Default 100. Do not change this unless its necessary.                                          |
| <code>bgEstimate</code>  | If FALSE skips background estimation from synonymous variants and uses predefined values estimated from COSMIC synonymous variants.                           |
| <code>ignoreGenes</code> | Ignore these genes from analysis. Default NULL. Helpful in case data contains large number of variants belonging to polymorphic genes such as mucins and TTN. |

## Details

This is the re-implimentation of algorithm defined in OncodriveCLUST article. Concept is based on the fact that most of the variants in cancer causing genes are enriched at few specific loci (aka hotspots). This method takes advantage of such positions to identify cancer genes. Cluster score of 1 means, a single hotspot hosts all observed variants. If you use this function, please cite OncodriveCLUST article.

## Value

data table of genes ordered according to p-values.

## References

Tamborero D, Gonzalez-Perez A and Lopez-Bigas N. OncodriveCLUST: exploiting the positional clustering of somatic mutations to identify cancer genes. *Bioinformatics*. 2013; doi: 10.1093/bioinformatics/btt395s

## See Also

[plotOncodrive](#)

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.sig <- oncodrive(maf = laml, AACol = 'Protein_Change', minMut = 5)
```

---

|                   |                                               |
|-------------------|-----------------------------------------------|
| OncogenicPathways | <i>Enrichment of known oncogenic pathways</i> |
|-------------------|-----------------------------------------------|

---

**Description**

Checks for enrichment of known oncogenic pathways

**Usage**

```
OncogenicPathways(maf, pathways = NULL, fontSize = 1, panelWidths = c(2, 4, 4))
```

**Arguments**

|             |                                                                                                                                                                      |
|-------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| maf         | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                                  |
| pathways    | Can be a two column data.frame/tsv-file with pathway-name and genes involved in them. Default 'NULL', uses a predefined list of pathways. See reference for details. |
| fontSize    | Default 1                                                                                                                                                            |
| panelWidths | Default c(2, 4, 4)                                                                                                                                                   |

**Details**

Oncogenic signalling pathways are derived from TCGA cohorts. See reference for details.

**Value**

Prints fraction of altered pathway

**References**

Sanchez-Vega F, Mina M, Armenia J, Chatila WK, Luna A, La KC, Dimitriadoy S, Liu DL, Kantheti HS, Saghafeinia S et al. 2018. Oncogenic Signaling Pathways in The Cancer Genome Atlas. Cell 173: 321-337 e310

**See Also**

[PlotOncogenicPathways](#)

**Examples**

```
lam1.maf <- system.file("extdata", "tcga_lam1.maf.gz", package = "maftools")
lam1 <- read.maf(maf = lam1.maf)
OncogenicPathways(maf = lam1)
```

---

oncoplot

*draw an oncoplot*


---

## Description

takes output generated by read.maf and draws an oncoplot

## Usage

```
oncoplot(
  maf,
  top = 20,
  minMut = NULL,
  genes = NULL,
  altered = FALSE,
  drawRowBar = TRUE,
  drawColBar = TRUE,
  leftBarData = NULL,
  leftBarLims = NULL,
  rightBarData = NULL,
  rightBarLims = NULL,
  topBarData = NULL,
  logColBar = FALSE,
  includeColBarCN = TRUE,
  clinicalFeatures = NULL,
  annotationColor = NULL,
  annotationDat = NULL,
  pathways = NULL,
  path_order = NULL,
  selectedPathways = NULL,
  pwLineCol = "#535c68",
  pwLineWd = 1,
  draw_titv = FALSE,
  titv_col = NULL,
  showTumorSampleBarcodes = FALSE,
  barcode_mar = 4,
  barcodeSrt = 90,
  gene_mar = 5,
  anno_height = 1,
  legend_height = 4,
  sortByAnnotation = FALSE,
  groupAnnotationBySize = TRUE,
  annotationOrder = NULL,
  sortByMutation = FALSE,
  keepGeneOrder = FALSE,
  GeneOrderSort = TRUE,
  sampleOrder = NULL,
```

```

    additionalFeature = NULL,
    additionalFeaturePch = 20,
    additionalFeatureCol = "gray70",
    additionalFeatureCex = 0.9,
    genesToIgnore = NULL,
    removeNonMutated = TRUE,
    fill = TRUE,
    cohortSize = NULL,
    colors = NULL,
    cBioPortal = FALSE,
    bgCol = "#CCCCCC",
    borderCol = "white",
    annoBorderCol = NA,
    numericAnnoCol = NULL,
    drawBox = FALSE,
    fontSize = 0.8,
    SampleNameFontSize = 1,
    titleFontSize = 1.5,
    legendFontSize = 1.2,
    annotationFontSize = 1.2,
    sepwd_genes = 0.5,
    sepwd_samples = 0.25,
    writeMatrix = FALSE,
    colbar_pathway = FALSE,
    showTitle = TRUE,
    titleText = NULL,
    showPct = TRUE
  )

```

## Arguments

|                           |                                                                                                                                                                                                                                  |
|---------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>maf</code>          | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                                                                                              |
| <code>top</code>          | how many top genes to be drawn. defaults to 20.                                                                                                                                                                                  |
| <code>minMut</code>       | draw all genes with 'min' number of mutations. Can be an integer or fraction (of samples mutated), Default NULL                                                                                                                  |
| <code>genes</code>        | Just draw oncoplot for these genes. Default NULL.                                                                                                                                                                                |
| <code>altered</code>      | Default FALSE. Chooses top genes based on mutation status. If TRUE chooses top genes based alterations (CNV or mutation).                                                                                                        |
| <code>drawRowBar</code>   | logical. Plots right barplot for each gene. Default TRUE.                                                                                                                                                                        |
| <code>drawColBar</code>   | logical plots top barplot for each sample. Default TRUE.                                                                                                                                                                         |
| <code>leftBarData</code>  | Data for leftside barplot. Must be a data.frame with two columns containing gene names and values. Default 'NULL'                                                                                                                |
| <code>leftBarLims</code>  | limits for 'leftBarData'. Default 'NULL'.                                                                                                                                                                                        |
| <code>rightBarData</code> | Data for rightside barplot. Must be a data.frame with two columns containing gene names and values. Default 'NULL' which draws distribution by variant classification. This option is applicable when only 'drawRowBar' is TRUE. |

|                         |                                                                                                                                                                                                                                                                                                  |
|-------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| rightBarLims            | limits for 'rightBarData'. Default 'NULL'.                                                                                                                                                                                                                                                       |
| topBarData              | Default 'NULL' which draws absolute number of mutation load for each sample. Can be overridden by choosing one clinical indicator(Numeric) or by providing a two column data.frame containing sample names and values for each sample. This option is applicable when only 'drawColBar' is TRUE. |
| logColBar               | Plot top bar plot on log10 scale. Default FALSE.                                                                                                                                                                                                                                                 |
| includeColBarCN         | Whether to include CN in column bar plot. Default TRUE                                                                                                                                                                                                                                           |
| clinicalFeatures        | columns names from 'clinical.data' slot of MAF to be drawn in the plot. Default NULL.                                                                                                                                                                                                            |
| annotationColor         | Custom colors to use for 'clinicalFeatures'. Must be a named list containing a named vector of colors. Default NULL. See example for more info.                                                                                                                                                  |
| annotationDat           | If MAF file was read without clinical data, provide a custom data.frame with a column Tumor_Sample_Barcode containing sample names along with rest of columns with annotations. You can specify which columns to be drawn using 'clinicalFeatures' argument.                                     |
| pathways                | Default 'NULL'. Can be 'auto', or a two column data.frame/tsv-file with genes and corresponding pathway mappings.'                                                                                                                                                                               |
| path_order              | Default 'NULL' Manually specify the order of pathways                                                                                                                                                                                                                                            |
| selectedPathways        | Manually provide the subset of pathway names to be selected from 'pathways'. Default NULL. In case 'pathways' is 'auto' draws top 3 altered pathways.                                                                                                                                            |
| pwLineCol               | Color for the box around the pathways Default #535c68                                                                                                                                                                                                                                            |
| pwLineWd                | Line width for the box around the pathways Default Default 1                                                                                                                                                                                                                                     |
| draw_titv               | logical Includes TiTv plot. FALSE                                                                                                                                                                                                                                                                |
| titv_col                | named vector of colors for each transition and transversion classes. Should be of length six with the names "C>T" "C>G" "C>A" "T>A" "T>C" "T>G". Default NULL.                                                                                                                                   |
| showTumorSampleBarcodes | logical to include sample names.                                                                                                                                                                                                                                                                 |
| barcode_mar             | Margin width for sample names. Default 4                                                                                                                                                                                                                                                         |
| barcodeSrt              | Rotate sample labels. Default 90.                                                                                                                                                                                                                                                                |
| gene_mar                | Margin width for gene names. Default 5                                                                                                                                                                                                                                                           |
| anno_height             | Height of plotting area for sample annotations. Default 1                                                                                                                                                                                                                                        |
| legend_height           | Height of plotting area for legend. Default 4                                                                                                                                                                                                                                                    |
| sortByAnnotation        | logical sort oncomatrix (samples) by provided 'clinicalFeatures'. Sorts based on first 'clinicalFeatures'. Defaults to FALSE. column-sort                                                                                                                                                        |
| groupAnnotationBySize   | Further group 'sortByAnnotation' orders by their size. Defaults to TRUE. Largest groups comes first.                                                                                                                                                                                             |

|                      |                                                                                                                                                                   |
|----------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| annotationOrder      | Manually specify order for annotations. Works only for first 'clinicalFeatures'. Default NULL.                                                                    |
| sortByMutation       | Force sort matrix according mutations. Helpful in case of MAF was read along with copy number data. Default FALSE.                                                |
| keepGeneOrder        | logical whether to keep order of given genes. Default FALSE, order according to mutation frequency                                                                |
| GeneOrderSort        | logical this is applicable when 'keepGeneOrder' is TRUE. Default TRUE                                                                                             |
| sampleOrder          | Manually speify sample names for oncolplot ordering. Default NULL.                                                                                                |
| additionalFeature    | a vector of length two indicating column name in the MAF and the factor level to be highlighted. Provide a list of values for highlighting more than one features |
| additionalFeaturePch | Default 20                                                                                                                                                        |
| additionalFeatureCol | Default "gray70"                                                                                                                                                  |
| additionalFeatureCex | Default 0.9                                                                                                                                                       |
| genesToIgnore        | do not show these genes in Oncoplot. Default NULL.                                                                                                                |
| removeNonMutated     | Logical. If TRUE removes samples with no mutations in the oncoplot for better visualization. Default TRUE.                                                        |
| fill                 | Logical. If TRUE draws genes and samples as blank grids even when they are not altered.                                                                           |
| cohortSize           | Number of sequenced samples in the cohort. Default all samples from Cohort. You can manually specify the cohort size. Default NULL                                |
| colors               | named vector of colors for each Variant_Classification.                                                                                                           |
| cBioPortal           | Adds annotations similar to cBioPortals MutationMapper and collapse Variants into Truncating and rest.                                                            |
| bgCol                | Background grid color for wild-type (not-mutated) samples. Default gray - "#CCCCCC"                                                                               |
| borderCol            | border grid color (not-mutated) samples. Default 'white'.                                                                                                         |
| annoBorderCol        | border grid color for annotations. Default NA.                                                                                                                    |
| numericAnnoCol       | color palette used for numeric annotations. Default 'YlOrBr' from RColorBrewer                                                                                    |
| drawBox              | logical whether to draw a box around main matrix. Default FALSE                                                                                                   |
| fontSize             | font size for gene names. Default 0.8.                                                                                                                            |
| SampleNamefontSize   | font size for sample names. Default 1                                                                                                                             |
| titleFontSize        | font size for title. Default 1.5                                                                                                                                  |
| legendFontSize       | font size for legend. Default 1.2                                                                                                                                 |
| annotationFontSize   | font size for annotations. Default 1.2                                                                                                                            |

|                |                                                                            |
|----------------|----------------------------------------------------------------------------|
| sepwd_genes    | size of lines seperating genes. Default 0.5                                |
| sepwd_samples  | size of lines seperating samples. Default 0.25                             |
| writeMatrix    | writes character coded matrix used to generate the plot to an output file. |
| colbar_pathway | Draw top column bar with respect to diplayed pathway. Default FALSE.       |
| showTitle      | Default TRUE                                                               |
| titleText      | Custom title. Default 'NULL'                                               |
| showPct        | Default TRUE. Shows percent altered to the right side of the plot.         |

### Details

Takes maf file as input and plots it as a matrix. Any desired clincal features can be added at the bottom of the oncoplot by providing clinicalFeatures. Oncoplot can be sorted either by mutations or by clinicalFeatures using arguments sortByMutation and sortByAnnotation respectively.

### Value

None.

### See Also

[oncostrip](#)

### Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml.clin = system.file('extdata', 'tcga_laml_annot.tsv', package = 'maftools')
laml <- read.maf(maf = laml.maf, clinicalData = laml.clin)
#Basic oncoplot
oncoplot(maf = laml, top = 3)
#Changing colors for variant classifications (You can use any colors, here in this example we will use a color palette)
col = RColorBrewer::brewer.pal(n = 8, name = 'Paired')
names(col) = c('Frame_Shift_Del', 'Missense_Mutation', 'Nonsense_Mutation', 'Multi_Hit', 'Frame_Shift_Ins',
               'In_Frame_Ins', 'Splice_Site', 'In_Frame_Del')
#Color coding for FAB classification; try getAnnotations(x = laml) to see available annotations.
fabcolors = RColorBrewer::brewer.pal(n = 8, name = 'Spectral')
names(fabcolors) = c("M0", "M1", "M2", "M3", "M4", "M5", "M6", "M7")
fabcolors = list(FAB_classification = fabcolors)
oncoplot(maf = laml, colors = col, clinicalFeatures = 'FAB_classification', sortByAnnotation = TRUE, annotationCol = fabcolors)
```

---

oncostrip

*draw an oncostrip similar to cBioportal oncoprinter output.*

---

### Description

draw an oncostrip similar to cBioportal oncoprinter output.

**Usage**

```
oncostrip(maf = NULL, ...)
```

**Arguments**

|     |                                                                  |
|-----|------------------------------------------------------------------|
| maf | an <a href="#">MAF</a> object generated by <code>read.maf</code> |
| ... | arguments passed <a href="#">oncoplot</a>                        |

**Details**

This is just a wrapper around [oncoplot](#) with `drawRowBar` and `drawColBar` set to `FALSE`

**Value**

None.

**See Also**

[oncoplot](#)

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
dev.new()
oncostrip(maf = laml, genes = c('NPM1', 'RUNX1'))
```

---

pfamDomains

*pfam domain annotation and summarization.*

---

**Description**

Summarizes amino acid positions and annotates them with pfam domain information.

**Usage**

```
pfamDomains(  
  maf = NULL,  
  AACol = NULL,  
  summarizeBy = "AAPos",  
  top = 5,  
  domainsToLabel = NULL,  
  baseName = NULL,  
  varClass = "nonSyn",  
  width = 5,  
  height = 5,  
  labelSize = 1  
)
```

**Arguments**

|                |                                                                                          |
|----------------|------------------------------------------------------------------------------------------|
| maf            | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                      |
| AACol          | manually specify column name for amino acid changes. Default looks for field 'AChange'   |
| summarizeBy    | Summarize domains by amino acid position or conversions. Can be "AAPos" or "AChange"     |
| top            | How many top mutated domains to label in the scatter plot. Defaults to 5.                |
| domainsToLabel | Default NULL. Exclusive with top argument.                                               |
| baseName       | If given writes the results to output file. Default NULL.                                |
| varClass       | which variants to consider for summarization. Can be nonSyn, Syn or all. Default nonSyn. |
| width          | width of the file to be saved.                                                           |
| height         | height of the file to be saved.                                                          |
| labelSize      | font size for labels. Default 1.                                                         |

**Value**

returns a list two tables summarized by amino acid positions and domains respectively. Also plots top 5 most mutated domains as scatter plot.

**Examples**

```
lam1.maf <- system.file("extdata", "tcga_lam1.maf.gz", package = "maftools")
lam1 <- read.maf(maf = lam1.maf)
pfamDomains(maf = lam1, AACol = 'Protein_Change')
```

---

|                |                                                                                  |
|----------------|----------------------------------------------------------------------------------|
| plotApobecDiff | <i>Plot differences between APOBEC enriched and non-APOBEC enriched samples.</i> |
|----------------|----------------------------------------------------------------------------------|

---

**Description**

Plots differences between APOBEC enriched and non-APOBEC enriched samples

**Usage**

```
plotApobecDiff(
  tnm,
  maf,
  pVal = 0.05,
  title_size = 1,
  axis_lwd = 1,
  font_size = 1.2
)
```

## Arguments

|            |                                                           |
|------------|-----------------------------------------------------------|
| tnm        | output generated by <a href="#">trinucleotideMatrix</a>   |
| maf        | an <a href="#">MAF</a> object used to generate the matrix |
| pVal       | p-value threshold for fisher's test. Default 0.05.        |
| title_size | size of title. Default 1.3                                |
| axis_lwd   | axis width. Default 1                                     |
| font_size  | font size. Default 1.2                                    |

## Details

Plots differences between APOBEC enriched and non-APOBEC enriched samples (TCW). Plot includes differences in mutations load, tCw motif distribution and top genes altered.

## Value

list of table containing differentially altered genes. This can be passed to [forestPlot](#) to plot results.

## See Also

[trinucleotideMatrix](#) [plotSignatures](#)

## Examples

```
## Not run:
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.tnm <- trinucleotideMatrix(maf = laml, ref_genome = 'BSgenome.Hsapiens.UCSC.hg19', prefix = 'chr',
add = TRUE, useSyn = TRUE)
plotApobecDiff(laml.tnm)

## End(Not run)
```

---

plotCBSsegments

*Plots segmented copy number data.*

---

## Description

Plots segmented copy number data.

**Usage**

```
plotCBSsegments(
  cbsFile = NULL,
  maf = NULL,
  tsb = NULL,
  savePlot = FALSE,
  ylims = NULL,
  seg_size = 0.1,
  width = 6,
  height = 3,
  genes = NULL,
  ref.build = "hg19",
  writeTable = FALSE,
  removeXY = FALSE,
  color = NULL
)
```

**Arguments**

|            |                                                                                                                                                                                                                                                                                                 |
|------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| cbsFile    | CBS segmented copy number file. Column names should be Sample, Chromosome, Start, End, Num_Probes and Segment_Mean (log2 scale).                                                                                                                                                                |
| maf        | optional <a href="#">MAF</a>                                                                                                                                                                                                                                                                    |
| tsb        | If segmentation file contains many samples (as in gistic input), specify sample name here. Default plots head 1 sample. Set 'ALL' for plotting all samples. If you are mapping maf, make sure sample names in Sample column of segmentation file matches to those Tumor_Sample_Barcodes in MAF. |
| savePlot   | If true plot is saved as pdf.                                                                                                                                                                                                                                                                   |
| ylims      | Default NULL                                                                                                                                                                                                                                                                                    |
| seg_size   | Default 0.1                                                                                                                                                                                                                                                                                     |
| width      | width of plot                                                                                                                                                                                                                                                                                   |
| height     | height of plot                                                                                                                                                                                                                                                                                  |
| genes      | If given and maf object is specified, maps all mutations from maf onto segments. Default NULL                                                                                                                                                                                                   |
| ref.build  | Reference build for chromosome sizes. Can be hg18, hg19 or hg38. Default hg19.                                                                                                                                                                                                                  |
| writeTable | If true and if maf object is specified, writes plot data with each variant and its corresponding copynumber to an output file.                                                                                                                                                                  |
| removeXY   | don not plot sex chromosomes.                                                                                                                                                                                                                                                                   |
| color      | Manually specify color scheme for chromosomes. Default NULL. i.e, alternating Gray70 and midnightblue                                                                                                                                                                                           |

**Details**

this function takes segmented copy number data and plots it. If MAF object is specified, all mutations are highlighted on the plot.

**Value**

Draws plot

**Examples**

```
tcga.ab.009.seg <- system.file("extdata", "TCGA.AB.3009.hg19.seg.txt", package = "maftools")
plotCBSsegments(cbsFile = tcga.ab.009.seg)
```

---

|              |                                                    |
|--------------|----------------------------------------------------|
| plotClusters | <i>Plot density plots from cluterling results.</i> |
|--------------|----------------------------------------------------|

---

**Description**

Plots results from inferHeterogeneity.

**Usage**

```
plotClusters(  
  clusters,  
  tsb = NULL,  
  genes = NULL,  
  showCNvars = FALSE,  
  colors = NULL  
)
```

**Arguments**

|            |                                                                                                                                                           |
|------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------|
| clusters   | clustering results from <a href="#">inferHeterogeneity</a>                                                                                                |
| tsb        | sample to plot from clustering results. Default plots all samples from results.                                                                           |
| genes      | genes to highlight on the plot. Can be a vector of gene names, CN_altered to label copy number altered varinats, or all to label all genes. Default NULL. |
| showCNvars | show copy numbered altered variants on the plot. Default FALSE.                                                                                           |
| colors     | manual colors for clusters. Default NULL.                                                                                                                 |

**Value**

returns nothing.

**See Also**

[inferHeterogeneity](#)

Examples

```
## Not run:
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
seg = system.file('extdata', 'TCGA.AB.3009.hg19.seg.txt', package = 'maftools')
TCGA.AB.3009.clust <- inferHeterogeneity(maf = laml, tsb = 'TCGA-AB-3009',
segFile = seg, vafCol = 'i_TumorVAF_WU')
plotClusters(TCGA.AB.3009.clust, genes = c('NF1', 'SUZ12'), showCNvars = TRUE)

## End(Not run)
```

---

|                |                                                             |
|----------------|-------------------------------------------------------------|
| plotCophenetic | <i>Draw an elbow plot of cophenetic correlation metric.</i> |
|----------------|-------------------------------------------------------------|

---

Description

Draw an elbow plot of cophenetic correlation metric.

Usage

```
plotCophenetic(res = NULL, bestFit = NULL)
```

Arguments

|         |                                                |
|---------|------------------------------------------------|
| res     | output from <a href="#">estimateSignatures</a> |
| bestFit | rank to highlight. Default NULL                |

Details

This function draws an elbow plot of cophenetic correlation metric.

See Also

[estimateSignatures](#) [plotCophenetic](#)

---

|                       |                                                       |
|-----------------------|-------------------------------------------------------|
| plotEnrichmentResults | <i>Plots results from clinicalEnrichment analysis</i> |
|-----------------------|-------------------------------------------------------|

---

Description

Plots results from clinicalEnrichment analysis

**Usage**

```
plotEnrichmentResults(  
  enrich_res,  
  pVal = 0.05,  
  ORthr = 1,  
  featureLvls = NULL,  
  cols = NULL,  
  annoFontSize = 0.8,  
  geneFontSize = 0.8,  
  legendFontSize = 0.8,  
  showTitle = TRUE  
)
```

**Arguments**

|                |                                                                                             |
|----------------|---------------------------------------------------------------------------------------------|
| enrich_res     | results from <a href="#">clinicalEnrichment</a> or <a href="#">signatureEnrichment</a>      |
| pVal           | Default 0.05                                                                                |
| ORthr          | Default 1. Odds ratio threshold. >1 indicates positive enrichment in the group of interest. |
| featureLvls    | Plot results from the selected levels. Default NULL, plots all.                             |
| cols           | named vector of colors for factor in a clinical feature. Default NULL                       |
| annoFontSize   | cex for annotation font size. Default 0.8                                                   |
| geneFontSize   | cex for gene font size. Default 0.8                                                         |
| legendFontSize | cex for legend font size. Default 0.8                                                       |
| showTitle      | Default TRUE                                                                                |

**Value**

returns nothing.

**See Also**

[clinicalEnrichment](#) [signatureEnrichment](#)

---

|                |                           |
|----------------|---------------------------|
| plotmafSummary | <i>Plots maf summary.</i> |
|----------------|---------------------------|

---

**Description**

Plots maf summary.

**Usage**

```
plotmafSummary(
  maf,
  rmOutlier = TRUE,
  dashboard = TRUE,
  titvRaw = TRUE,
  log_scale = FALSE,
  addStat = NULL,
  showBarcodes = FALSE,
  fs = 1,
  textSize = 0.8,
  color = NULL,
  titleSize = c(1, 0.8),
  titvColor = NULL,
  top = 10
)
```

**Arguments**

|              |                                                                                                         |
|--------------|---------------------------------------------------------------------------------------------------------|
| maf          | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                     |
| rmOutlier    | If TRUE removes outlier from boxplot.                                                                   |
| dashboard    | If FALSE plots simple summary instead of dashboard style.                                               |
| titvRaw      | TRUE. If false instead of raw counts, plots fraction.                                                   |
| log_scale    | FALSE. If TRUE log10 transforms Variant Classification, Variant Type and Variants per sample sub-plots. |
| addStat      | Can be either mean or median. Default NULL.                                                             |
| showBarcodes | include sample names in the top bar plot.                                                               |
| fs           | base size for text. Default 1                                                                           |
| textSize     | font size if showBarcodes is TRUE. Default 0.8                                                          |
| color        | named vector of colors for each Variant_Classification.                                                 |
| titleSize    | font size for title and subtitle. Default c(10, 8)                                                      |
| titvColor    | colors for SNV classifications.                                                                         |
| top          | include top n genes dashboard plot. Default 10.                                                         |

**Value**

Prints plot.

**See Also**

[read.maf](#) [MAF](#)

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf, useAll = FALSE)
plotmafSummary(maf = laml, addStat = 'median')
```

---

|              |                                                                |
|--------------|----------------------------------------------------------------|
| plotMosdepth | <i>Plot results from mosdepth output for Tumor/Normal pair</i> |
|--------------|----------------------------------------------------------------|

---

### Description

Plot results from mosdepth output for Tumor/Normal pair

### Usage

```
plotMosdepth(
  t_bed = NULL,
  n_bed = NULL,
  segment = TRUE,
  sample_name = NULL,
  col = c("#95a5a6", "#7f8c8d")
)
```

### Arguments

|             |                                          |
|-------------|------------------------------------------|
| t_bed       | mosdepth output from tumor               |
| n_bed       | mosdepth output from matched normal      |
| segment     | Performs CBS segmentation. Default TRUE  |
| sample_name | sample name. Default parses from 't_bed' |
| col         | Colors. Default c("#95a5a6", "#7f8c8d")  |

### Value

Invisibly returns [DNAcopy](#) object if 'segment' is 'TRUE'

### References

Pedersen BS, Quinlan AR. Mosdepth: quick coverage calculation for genomes and exomes. *Bioinformatics*. 2018;34(5):867-868. doi:10.1093/bioinformatics/btx699

---

|                |                                          |
|----------------|------------------------------------------|
| plotMosdepth_t | <i>Plot results from mosdepth output</i> |
|----------------|------------------------------------------|

---

### Description

Plot results from mosdepth output

**Usage**

```
plotMosdepth_t(
  bed = NULL,
  col = c("#95a5a6", "#7f8c8d"),
  sample_name = NULL,
  segment = FALSE
)
```

**Arguments**

|             |                                          |
|-------------|------------------------------------------|
| bed         | mosdepth output                          |
| col         | Colors. Default c("#95a5a6", "#7f8c8d")  |
| sample_name | sample name. Default parses from 'bed'   |
| segment     | Performs CBS segmentation. Default FALSE |

**Value**

Invisibly returns [DNAcopy](#) object if 'segment' is 'TRUE'

**References**

Pedersen BS, Quinlan AR. Mosdepth: quick coverage calculation for genomes and exomes. *Bioinformatics*. 2018;34(5):867-868. doi:10.1093/bioinformatics/btx699

---

|               |                                     |
|---------------|-------------------------------------|
| plotOncodrive | <i>Plots results from oncodrive</i> |
|---------------|-------------------------------------|

---

**Description**

Takes results from oncodrive and plots them as a scatter plot. Size of the gene shows number of clusters (hotspots), x-axis can either be an absolute number of variants accumulated in these clusters or a fraction of total variants found in these clusters. y-axis is fdr values transformed into -log10 for better representation. Labels indicate Gene name with number clusters observed.

**Usage**

```
plotOncodrive(
  res = NULL,
  fdrCutOff = 0.05,
  useFraction = FALSE,
  colCode = NULL,
  bubbleSize = 1,
  labelSize = 1
)
```

**Arguments**

|             |                                                                                       |
|-------------|---------------------------------------------------------------------------------------|
| res         | results from <a href="#">oncodrive</a>                                                |
| fdrCutoff   | fdr cutoff to call a gene as a driver.                                                |
| useFraction | if TRUE uses a fraction of total variants as X-axis scale instead of absolute counts. |
| colCode     | Colors to use for indicating significant and non-significant genes. Default NULL      |
| bubbleSize  | Size for bubbles. Default 2.                                                          |
| labelSize   | font size for labelling genes. Default 1.                                             |

**Value**

Nothing

**See Also**

[oncodrive](#)

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.sig <- oncodrive(maf = laml, AACol = 'Protein_Change', minMut = 5)
plotOncodrive(res = laml.sig, fdrCutoff = 0.1)
```

---

PlotOncogenicPathways *Plot oncogenic pathways*

---

**Description**

Plot oncogenic pathways

**Usage**

```
PlotOncogenicPathways(
  maf,
  pathways = NULL,
  fullPathway = FALSE,
  removeNonMutated = TRUE,
  tsgCol = "red",
  ogCol = "royalblue",
  fontSize = 0.6,
  showTumorSampleBarcodes = FALSE,
  sampleOrder = NULL,
  SampleNamefontSize = 0.6
)
```

Arguments

|                         |                                                                            |
|-------------------------|----------------------------------------------------------------------------|
| maf                     | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>        |
| pathways                | Name of pathways to be drawn                                               |
| fullPathway             | Include all genes from the pathway. Default FALSE only plots mutated genes |
| removeNonMutated        | Default TRUE                                                               |
| tsgCol                  | Color for tumor suppressor genes. Default red                              |
| ogCol                   | Color for onco genes. Default royalblue                                    |
| fontSize                | Default 0.6                                                                |
| showTumorSampleBarcodes | logical to include sample names.                                           |
| sampleOrder             | Manually specify sample names for oncoplot ordering. Default NULL.         |
| SampleNamefontSize      | font size for sample names. Default 10                                     |

Details

Draws oncoplot of oncogenic pathway.

References

Sanchez-Vega F, Mina M, Armenia J, Chatila WK, Luna A, La KC, Dimitriadou S, Liu DL, Kantheti HS, Saghafeini S et al. 2018. Oncogenic Signaling Pathways in The Cancer Genome Atlas. Cell 173: 321-337 e310

See Also

[OncogenicPathways](#)

Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
PlotOncogenicPathways(maf = laml, pathways = "RTK-RAS")
```

---

|             |                                |
|-------------|--------------------------------|
| plotProtein | <i>Display protein domains</i> |
|-------------|--------------------------------|

---

Description

Display protein domains

**Usage**

```
plotProtein(
  gene,
  refSeqID = NULL,
  proteinID = NULL,
  domainAlpha = 0.9,
  showLegend = FALSE,
  bgBorderCol = "black",
  axisTextSize = c(1, 1),
  roundedRect = TRUE,
  domainBorderCol = "black",
  showDomainLabel = TRUE,
  domainLabelSize = 0.8,
  titleSize = c(1.2, 1),
  legendTxtSize = 1,
  legendNcol = 1
)
```

**Arguments**

|                 |                                                                                         |
|-----------------|-----------------------------------------------------------------------------------------|
| gene            | HGNC symbol for which protein structure to be drawn.                                    |
| refSeqID        | RefSeq transcript identifier for gene if known.                                         |
| proteinID       | RefSeq protein identifier for gene if known.                                            |
| domainAlpha     | Default 1                                                                               |
| showLegend      | Default TRUE                                                                            |
| bgBorderCol     | Default "black". Set to NA to remove.                                                   |
| axisTextSize    | text size x and y tick labels. Default c(1,1).                                          |
| roundedRect     | Default TRUE. If 'TRUE' domains are drawn with rounded corners. Requires berryFunctions |
| domainBorderCol | Default "black". Set to NA to remove.                                                   |
| showDomainLabel | Default TRUE                                                                            |
| domainLabelSize | text size for domain labels. Default 0.8                                                |
| titleSize       | font size for title and subtitle. Default c(1.2, 1)                                     |
| legendTxtSize   | Text size for legend. Default 0.8                                                       |
| legendNcol      | Default 1                                                                               |

**Examples**

```
par(mfrow = c(2, 1))
plotProtein(gene = "KIT")
plotProtein(gene = "DNMT3A")
```

---

|                |                                               |
|----------------|-----------------------------------------------|
| plotSignatures | <i>Plots decomposed mutational signatures</i> |
|----------------|-----------------------------------------------|

---

## Description

Takes results from [extractSignatures](#) and plots decomposed mutational signatures as a barplot.

## Usage

```
plotSignatures(
  nmfRes = NULL,
  contributions = FALSE,
  color = NULL,
  patient_order = NULL,
  font_size = 1.2,
  show_title = TRUE,
  sig_db = "legacy",
  axis_lwd = 2,
  title_size = 0.9,
  show_barcodes = FALSE,
  yaxisLim = 0.3,
  ...
)
```

## Arguments

|               |                                                                             |
|---------------|-----------------------------------------------------------------------------|
| nmfRes        | results from <a href="#">extractSignatures</a>                              |
| contributions | If TRUE plots contribution of signatures in each sample.                    |
| color         | colors for each Ti/Tv conversion class. Default NULL                        |
| patient_order | User defined ordering of samples. Default NULL.                             |
| font_size     | font size. Default 1.2                                                      |
| show_title    | If TRUE compares signatures to COSMIC signatures and prints them as title   |
| sig_db        | Only applicable if show_title is TRUE. Can be legacy or SBS. Default legacy |
| axis_lwd      | axis width. Default 2.                                                      |
| title_size    | size of title. Default 1.3                                                  |
| show_barcodes | Default FALSE                                                               |
| yaxisLim      | Default 0.3. If NA autoscales.                                              |
| ...           | further plot options passed to <a href="#">barplot</a>                      |

## Value

Nothing

## See Also

[trinucleotideMatrix](#) [plotSignatures](#)

plotTiTv

*Plot Transition and Trasnversion ratios.***Description**

Takes results generated from `titv` and plots the Ti/Tv ratios and contributions of 6 mutational conversion classes in each sample.

**Usage**

```
plotTiTv(
  res = NULL,
  plotType = "both",
  sampleOrder = NULL,
  color = NULL,
  showBarcodes = FALSE,
  textSize = 0.8,
  baseFontSize = 1,
  axisTextSize = c(1, 1),
  plotNotch = FALSE
)
```

**Arguments**

|                           |                                                                   |
|---------------------------|-------------------------------------------------------------------|
| <code>res</code>          | results generated by <code>titv</code>                            |
| <code>plotType</code>     | Can be 'bar', 'box' or 'both'. Defaults to 'both'                 |
| <code>sampleOrder</code>  | Sample names in which the barplot should be ordered. Default NULL |
| <code>color</code>        | named vector of colors for each conversion class.                 |
| <code>showBarcodes</code> | Whether to include sample names for barplot                       |
| <code>textSize</code>     | fontsize if showBarcodes is TRUE. Deafult 2.                      |
| <code>baseFontSize</code> | font size. Deafult 1.                                             |
| <code>axisTextSize</code> | text size x and y tick labels. Default c(1,1).                    |
| <code>plotNotch</code>    | logical. Include notch in boxplot.                                |

**Value**

None.

**See Also**

`titv`

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.titv = titv(maf = laml, useSyn = TRUE)
plotTiTv(laml.titv)
```

---

plotVaf

*Plots vaf distribution of genes*

---

## Description

Plots vaf distribution of genes as a boxplot. Each dot in the jitter is a variant.

## Usage

```
plotVaf(
  maf,
  vafCol = NULL,
  genes = NULL,
  top = 10,
  orderByMedian = TRUE,
  keepGeneOrder = FALSE,
  flip = FALSE,
  fn = NULL,
  gene_fs = 0.8,
  axis_fs = 0.8,
  height = 5,
  width = 5,
  showN = TRUE,
  color = NULL
)
```

## Arguments

|               |                                                                         |
|---------------|-------------------------------------------------------------------------|
| maf           | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>     |
| vafCol        | manually specify column name for vafs. Default looks for column 't_vaf' |
| genes         | specify genes for which plots has to be generated                       |
| top           | if genes is NULL plots top n number of genes. Defaults to 5.            |
| orderByMedian | Orders genes by decreasing median VAF. Default TRUE                     |
| keepGeneOrder | keep gene order. Default FALSE                                          |
| flip          | if TRUE, flips axes. Default FALSE                                      |
| fn            | Filename. If given saves plot as a output pdf. Default NULL.            |
| gene_fs       | font size for gene names. Default 0.8                                   |

|         |                                          |
|---------|------------------------------------------|
| axis_fs | font size for axis. Default 0.8          |
| height  | Height of plot to be saved. Default 5    |
| width   | Width of plot to be saved. Default 4     |
| showN   | if TRUE, includes number of observations |
| color   | manual colors. Default NULL.             |

**Value**

Nothing.

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
plotVaf(maf = laml, vafCol = 'i_TumorVAF_WU')
```

---

|               |                                               |
|---------------|-----------------------------------------------|
| prepareMutSig | <i>Prepares MAF file for MutSig analysis.</i> |
|---------------|-----------------------------------------------|

---

**Description**

Corrects gene names for MutSig compatibility.

**Usage**

```
prepareMutSig(maf, fn = NULL)
```

**Arguments**

|     |                                                                                             |
|-----|---------------------------------------------------------------------------------------------|
| maf | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                         |
| fn  | basename for output file. If provided writes MAF to an output file with the given basename. |

**Details**

MutSig/MutSigCV is most widely used program for detecting driver genes. However, we have observed that covariates files (gene.covariates.txt and exome\_full192.coverage.txt) which are bundled with MutSig have non-standard gene names (non Hugo\_Symbols). This discrepancy between Hugo\_Symbols in MAF and non-Hugo\_symbols in covariates file causes MutSig program to ignore such genes. For example, KMT2D - a well known driver gene in Esophageal Carcinoma is represented as MLL2 in MutSig covariates. This causes KMT2D to be ignored from analysis and is represented as an insignificant gene in MutSig results. This function attempts to correct such gene symbols with a manually curated list of gene names compatible with MutSig covariates list.

**Value**

returns a MAF with gene symbols corrected.

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
prepareMutSig(maf = laml)
```

---

prepAscat

---

*Prepare input files for ASCAT*


---

**Description**

Function takes the output from [gtMarkers](#) and generates ‘logR’ and ‘BAF’ files required for ASCAT analysis.

**Usage**

```
prepAscat(
  t_counts = NULL,
  n_counts = NULL,
  sample_name = NA,
  min_depth = 15,
  normalize = TRUE
)
```

**Arguments**

|             |                                                                                              |
|-------------|----------------------------------------------------------------------------------------------|
| t_counts    | read counts from tumor generated by <a href="#">gtMarkers</a>                                |
| n_counts    | read counts from normal generated by <a href="#">gtMarkers</a>                               |
| sample_name | Sample name. Used as a basename for output files. Default ‘NA’, parses from ‘t_counts’ file. |
| min_depth   | Min read depth required to consider a marker. Default 15                                     |
| normalize   | If TRUE, normalizes for library size. Default TRUE                                           |

**Details**

The function will filter SNPs with low coverage (default <15), estimate BAF, logR, and generates the input files for ASCAT. Alternatively, logR file can be segmented with [segmentLogR](#)

**References**

Van Loo P, Nordgard SH, Lingjærde OC, et al. Allele-specific copy number analysis of tumors. Proc Natl Acad Sci U S A. 2010;107(39):16910-16915. doi:10.1073/pnas.1009843107

**See Also**

[gtMarkers](#) [prepAscat\\_t](#) [segmentLogR](#)

---

|             |                                                         |
|-------------|---------------------------------------------------------|
| prepAscat_t | <i>Prepare input files for ASCAT tumor only samples</i> |
|-------------|---------------------------------------------------------|

---

## Description

Function takes the output from [gtMarkers](#) and generates 'logR' and 'BAF' files required for ASCAT analysis.

## Usage

```
prepAscat_t(t_counts = NULL, sample_name = NA, min_depth = 15)
```

## Arguments

|             |                                                                                            |
|-------------|--------------------------------------------------------------------------------------------|
| t_counts    | read counts from tumor generated by <a href="#">gtMarkers</a>                              |
| sample_name | Sample name. Used as a basename for output files. Default NA, parses from 't_counts' file. |
| min_depth   | Min read depth required to consider a marker. Default 15                                   |

## Details

The function will filter SNPs with low coverage (default <15), estimate BAF, logR, and generates the input files for ASCAT. Tumor 'logR' file will be normalized for median depth of coverage. Alternatively, logR file can be segmented with [segmentLogR](#)

## Value

Generates logR and BAF files required by ASCAT

## References

Van Loo P, Nordgard SH, Lingjærde OC, et al. Allele-specific copy number analysis of tumors. Proc Natl Acad Sci U S A. 2010;107(39):16910-16915. doi:10.1073/pnas.1009843107

## See Also

[gtMarkers](#) [prepAscat](#) [segmentLogR](#)

---

|              |                                                                |
|--------------|----------------------------------------------------------------|
| rainfallPlot | <i>Rainfall plot to display hyper mutated genomic regions.</i> |
|--------------|----------------------------------------------------------------|

---

## Description

Plots inter variant distance as a function of genomic locus.

## Usage

```
rainfallPlot(
  maf,
  tsb = NULL,
  detectChangePoints = FALSE,
  ref.build = "hg19",
  color = NULL,
  savePlot = FALSE,
  width = 6,
  height = 3,
  fontSize = 1.2,
  pointSize = 0.4
)
```

## Arguments

|                    |                                                                                                                                  |
|--------------------|----------------------------------------------------------------------------------------------------------------------------------|
| maf                | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a> . Required.                                                  |
| tsb                | specify sample names (Tumor_Sample_Barcodes) for which plotting has to be done. If NULL, draws plot for most mutated sample.     |
| detectChangePoints | If TRUE, detects genomic change points where potential kataegis are formed. Results are written to an output tab delimited file. |
| ref.build          | Reference build for chromosome sizes. Can be hg18, hg19 or hg38. Default hg19.                                                   |
| color              | named vector of colors for each conversion class.                                                                                |
| savePlot           | If TRUE plot is saved to output pdf. Default FALSE.                                                                              |
| width              | width of plot to be saved.                                                                                                       |
| height             | height of plot to be saved.                                                                                                      |
| fontSize           | Default 12.                                                                                                                      |
| pointSize          | Default 0.8.                                                                                                                     |

## Details

If ‘detectChangePoints’ is set to TRUE, this function will identify Kataegis loci. Kategis detection algorithm by Moritz Goretzky at WWU Munster, which exploits the definition of Kategis (six consecutive mutations with an avg. distance of 1000bp ) to identify hyper mutated genomic loci.

Algorithm starts with a double-ended queue to which six consecutive mutations are added and their average intermutation distance is calculated. If the average intermutation distance is larger than 1000, one element is added at the back of the queue and one is removed from the front. If the average intermutation distance is less or equal to 1000, further mutations are added until the average intermutation distance is larger than 1000. After that all mutations in the double-ended queue are written into output as one kataegis and the double-ended queue is reinitialized with six mutations.

## Value

Results are written to an output file with suffix changePoints.tsv

---

|          |                        |
|----------|------------------------|
| read.maf | <i>Read MAF files.</i> |
|----------|------------------------|

---

## Description

Takes tab delimited MAF (can be plain text or gz compressed) file as an input and summarizes it in various ways. Also creates oncomatrix - helpful for visualization.

## Usage

```
read.maf(
  maf,
  clinicalData = NULL,
  rmFlags = FALSE,
  removeDuplicatedVariants = TRUE,
  useAll = TRUE,
  gisticAllLesionsFile = NULL,
  gisticAmpGenesFile = NULL,
  gisticDelGenesFile = NULL,
  gisticScoresFile = NULL,
  cnLevel = "all",
  cnTable = NULL,
  isTCGA = FALSE,
  vc_nonSyn = NULL,
  verbose = TRUE
)
```

## Arguments

|              |                                                                                                                                             |
|--------------|---------------------------------------------------------------------------------------------------------------------------------------------|
| maf          | tab delimited MAF file. File can also be gz compressed. Required. Alternatively, you can also provide already read MAF file as a dataframe. |
| clinicalData | Clinical data associated with each sample/Tumor_Sample_Barcode in MAF. Could be a text file or a data.frame. Default NULL.                  |
| rmFlags      | Default FALSE. Can be TRUE or an integer. If TRUE removes all the top 20 FLAG genes. If integer, remove top n FLAG genes.                   |

|                                       |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |
|---------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>removeDuplicatedVariants</code> | removes repeated variants in a particular sample, mapped to multiple transcripts of same Gene. See Description. Default TRUE.                                                                                                                                                                                                                                                                                                                                                                            |
| <code>useAll</code>                   | logical. Whether to use all variants irrespective of values in <code>Mutation_Status</code> . Defaults to TRUE. If FALSE, only uses with values Somatic.                                                                                                                                                                                                                                                                                                                                                 |
| <code>gisticAllLesionsFile</code>     | All Lesions file generated by gistic. e.g; all_lesions.conf_XX.txt, where XX is the confidence level. Default NULL.                                                                                                                                                                                                                                                                                                                                                                                      |
| <code>gisticAmpGenesFile</code>       | Amplification Genes file generated by gistic. e.g; amp_genes.conf_XX.txt, where XX is the confidence level. Default NULL.                                                                                                                                                                                                                                                                                                                                                                                |
| <code>gisticDelGenesFile</code>       | Deletion Genes file generated by gistic. e.g; del_genes.conf_XX.txt, where XX is the confidence level. Default NULL.                                                                                                                                                                                                                                                                                                                                                                                     |
| <code>gisticScoresFile</code>         | scores.gistic file generated by gistic. Default NULL                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
| <code>cnLevel</code>                  | level of CN changes to use. Can be 'all', 'deep' or 'shallow'. Default uses all i.e, genes with both 'shallow' or 'deep' CN changes                                                                                                                                                                                                                                                                                                                                                                      |
| <code>cnTable</code>                  | Custom copynumber data if gistic results are not available. Input file or a data.frame should contain three columns in aforementioned order with gene name, Sample name and copy number status (either 'Amp' or 'Del'). Default NULL.                                                                                                                                                                                                                                                                    |
| <code>isTCGA</code>                   | Is input MAF file from TCGA source. If TRUE uses only first 12 characters from Tumor_Sample_Barcode.                                                                                                                                                                                                                                                                                                                                                                                                     |
| <code>vc_nonSyn</code>                | NULL. Provide manual list of variant classifications to be considered as non-synonymous. Rest will be considered as silent variants. Default uses Variant Classifications with High/Moderate variant consequences. <a href="https://m.ensembl.org/info/genome/variation/p">https://m.ensembl.org/info/genome/variation/p</a> "Frame_Shift_Del", "Frame_Shift_Ins", "Splice_Site", "Translation_Start_Site", "Nonsense_Mutation", "Nonstop_Mutation", "In_Frame_Del", "In_Frame_Ins", "Missense_Mutation" |
| <code>verbose</code>                  | TRUE logical. Default to be talkative and prints summary.                                                                                                                                                                                                                                                                                                                                                                                                                                                |

## Details

This function takes MAF file as input and summarizes them. If copy number data is available, e.g from GISTIC, it can be provided too via arguments `gisticAllLesionsFile`, `gisticAmpGenesFile`, and `gisticDelGenesFile`. Copy number data can also be provided as a custom table containing Gene name, Sample name and Copy Number status.

Note that if input MAF file contains multiple affected transcripts of a variant, this function by default removes them as duplicates, while keeping single unique entry per variant per sample. If you wish to keep all of them, set `removeDuplicatedVariants` to FALSE.

FLAGS - If you get a note on possible FLAGS while reading MAF, it means some of the top mutated genes are fishy. These genes are often non-pathogenic and passengers, but are frequently mutated in most of the public exome studies. Examples of such genes include TTN, MUC16, etc. This note can be ignored without any harm, it's only generated as to make user aware of such genes. See references for details on FLAGS.

**Value**

An object of class MAF.

**References**

Shyr C, Tarailo-Graovac M, Gottlieb M, Lee JJ, van Karnebeek C, Wasserman WW. FLAGS, frequently mutated genes in public exomes. BMC Med Genomics 2014; 7: 64.

**See Also**

[plotmafSummary](#) [write.mafSummary](#)

**Examples**

```
laml.maf = system.file("extdata", "tcga_laml.maf.gz", package = "maftools") #MAF file
laml.clin = system.file('extdata', 'tcga_laml_annot.tsv', package = 'maftools') #clinical data
laml = read.maf(maf = laml.maf, clinicalData = laml.clin)
```

---

readGistic

*Read and summarize gistic output.*


---

**Description**

A little function to summarize gistic output files. Summarized output is returned as a list of tables.

**Usage**

```
readGistic(
  gisticAllLesionsFile = NULL,
  gisticAmpGenesFile = NULL,
  gisticDelGenesFile = NULL,
  gisticScoresFile = NULL,
  cnLevel = "all",
  isTCGA = FALSE,
  verbose = TRUE
)
```

**Arguments**

**gisticAllLesionsFile**

All Lesions file generated by gistic. e.g; all\_lesions.conf\_XX.txt, where XX is the confidence level. Required. Default NULL.

**gisticAmpGenesFile**

Amplification Genes file generated by gistic. e.g; amp\_genes.conf\_XX.txt, where XX is the confidence level. Default NULL.

**gisticDelGenesFile**

Deletion Genes file generated by gistic. e.g; del\_genes.conf\_XX.txt, where XX is the confidence level. Default NULL.

|                  |                                                                                                                                     |
|------------------|-------------------------------------------------------------------------------------------------------------------------------------|
| gisticScoresFile | scores.gistic file generated by gistic.                                                                                             |
| cnLevel          | level of CN changes to use. Can be 'all', 'deep' or 'shallow'. Default uses all i.e, genes with both 'shallow' or 'deep' CN changes |
| isTCGA           | Is the data from TCGA. Default FALSE.                                                                                               |
| verbose          | Default TRUE                                                                                                                        |

**Details**

Requires output files generated from GISTIC. Gistic documentation can be found here <ftp://ftp.broadinstitute.org/pub/GISTIC>

**Value**

A list of summarized data.

**Examples**

```
all.lesions <- system.file("extdata", "all_lesions.conf_99.txt", package = "maftools")
amp.genes <- system.file("extdata", "amp_genes.conf_99.txt", package = "maftools")
del.genes <- system.file("extdata", "del_genes.conf_99.txt", package = "maftools")
scores.gistic <- system.file("extdata", "scores.gistic", package = "maftools")
laml.gistic = readGistic(gisticAllLesionsFile = all.lesions, gisticAmpGenesFile = amp.genes, gisticDelGenesFile =
```

---

|             |                                               |
|-------------|-----------------------------------------------|
| sampleSwaps | <i>Identify sample swaps and similarities</i> |
|-------------|-----------------------------------------------|

---

**Description**

Given a list BAM files, the function genotypes known SNPs and identifies potentially related samples. For the source of SNPs, see reference

**Usage**

```
sampleSwaps(
  bams = NULL,
  build = "hg19",
  prefix = NULL,
  add = TRUE,
  min_depth = 30,
  ncores = 4,
  ...
)
```

**Arguments**

|           |                                                                                                                             |
|-----------|-----------------------------------------------------------------------------------------------------------------------------|
| bams      | Input bam files. Required.                                                                                                  |
| build     | reference genome build. Default "hg19". Can be hg19 or hg38                                                                 |
| prefix    | Prefix to add or remove from contig names in SNP file. If BAM files are aligned GRCh37/38 genome, use prefix 'chr' to 'add' |
| add       | If prefix is used, default is to add prefix to contig names in SNP file. If FALSE prefix will be removed from contig names. |
| min_depth | Minimum read depth for a SNP to be considered. Default 30.                                                                  |
| ncores    | Default 4. Each BAM file will be launched on a separate thread. Works only on Unix and macOS.                               |
| ...       | Additional arguments passed to <a href="#">bamreadcounts</a>                                                                |

**Value**

a list with results summarized

**References**

Westphal, M., Frankhouser, D., Sonzone, C. et al. SMaSH: Sample matching using SNPs in humans. BMC Genomics 20, 1001 (2019). <https://doi.org/10.1186/s12864-019-6332-7>

---

segmentLogR

---

*Segment and plot log ratio values with DNACopy*


---

**Description**

The function takes logR file generated by [prepAscat](#) or [prepAscat\\_t](#) and performs segmentation with [DNACopy](#)

**Usage**

```
segmentLogR(tumor_logR = NULL, sample_name = NULL, build = "hg19")
```

**Arguments**

|             |                                                                                     |
|-------------|-------------------------------------------------------------------------------------|
| tumor_logR  | logR.txt file generated by <a href="#">prepAscat</a> or <a href="#">prepAscat_t</a> |
| sample_name | Default NULL. Parses from 'tumor_logR' file                                         |
| build       | Reference genome. Default hg19. Can be hg18, hg19, or hg38                          |

**Value**

Invisibly returns [DNACopy](#) object

**See Also**

[gtMarkers](#) [prepAscat](#)

---

|            |                                       |
|------------|---------------------------------------|
| setdiffMAF | <i>Set Operations for MAF objects</i> |
|------------|---------------------------------------|

---

**Description**

Set Operations for MAF objects

**Usage**

```
setdiffMAF(x, y, mafObj = TRUE, refAltMatch = TRUE, ...)

intersectMAF(x, y, refAltMatch = TRUE, mafObj = TRUE, ...)
```

**Arguments**

- x                    the first ‘MAF’ object.
- y                    the second ‘MAF’ object.
- mafObj              Return output as an ‘MAF’ object. Default ‘TRUE’
- refAltMatch        Set operations are done by matching ref and alt alleles in addition to loci (Default). If FALSE only loci (chr, start, end positions) are matched.
- ...                  other parameters passing to ‘subsetMaf’ for subsetting operations.

**Value**

subset table or an object of class **MAF-class**. If no overlaps found returns ‘NULL’

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
x <- subsetMaf(maf = laml, tsb = c('TCGA-AB-3009'))
y <- subsetMaf(maf = laml, tsb = c('TCGA-AB-2933'))
setdiffMAF(x, y)
intersectMAF(x, y) #Should return NULL due to no common variants
```

---

|                     |                                                                                                |
|---------------------|------------------------------------------------------------------------------------------------|
| signatureEnrichment | <i>Performs sample stratification based on signature contribution and enrichment analysis.</i> |
|---------------------|------------------------------------------------------------------------------------------------|

---

**Description**

Performs k-means clustering to assign signature to samples and performs enrichment analysis. Note  
- Do not use this function. This will be removed in future updates.

**Usage**

```
signatureEnrichment(maf, sig_res, minMut = 5, useCNV = FALSE, fn = NULL)
```

**Arguments**

|         |                                                                                                                                 |
|---------|---------------------------------------------------------------------------------------------------------------------------------|
| maf     | an <a href="#">MAF</a> object used for signature analysis.                                                                      |
| sig_res | Signature results from <a href="#">extractSignatures</a>                                                                        |
| minMut  | Consider only genes with minimum this number of samples mutated. Default 5.                                                     |
| useCNV  | whether to include copy number events. Only applicable when MAF is read along with copy number data. Default TRUE if available. |
| fn      | basename for output file. Default NULL.                                                                                         |

**Value**

result list containing p-values

**See Also**

[plotEnrichmentResults](#)

---

|                     |                                                                                      |
|---------------------|--------------------------------------------------------------------------------------|
| somaticInteractions | <i>Exact tests to detect mutually exclusive, co-occurring and altered gene-sets.</i> |
|---------------------|--------------------------------------------------------------------------------------|

---

**Description**

Performs Pair-wise Fisher's Exact test to detect mutually exclusive or co-occurring events.

**Usage**

```
somaticInteractions(
  maf,
  top = 25,
  genes = NULL,
  pvalue = c(0.05, 0.01),
  returnAll = TRUE,
  geneOrder = NULL,
  fontSize = 0.8,
  showSigSymbols = TRUE,
  showCounts = FALSE,
  countStats = "all",
  countType = "all",
  countsFontSize = 0.8,
  countsFontColor = "black",
  colPal = "BrBG",
  showSum = TRUE,
```

```

plotPadj = FALSE,
colNC = 9,
nShiftSymbols = 5,
sigSymbolsSize = 2,
sigSymbolsFontSize = 0.9,
pvSymbols = c(46, 42),
limitColorBreaks = TRUE
)

```

## Arguments

|                    |                                                                                                                             |
|--------------------|-----------------------------------------------------------------------------------------------------------------------------|
| maf                | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                         |
| top                | check for interactions among top 'n' number of genes. Defaults to top 25. genes                                             |
| genes              | List of genes among which interactions should be tested. If not provided, test will be performed between top 25 genes.      |
| pvalue             | Default c(0.05, 0.01) p-value threshold. You can provide two values for upper and lower threshold.                          |
| returnAll          | If TRUE returns test statistics for all pair of tested genes. Default FALSE, returns for only genes below pvalue threshold. |
| geneOrder          | Plot the results in given order. Default NULL.                                                                              |
| fontSize           | cex for gene names. Default 0.8                                                                                             |
| showSigSymbols     | Default TRUE. Highlight significant pairs                                                                                   |
| showCounts         | Default TRUE. Include number of events in the plot                                                                          |
| countStats         | Default 'all'. Can be 'all' or 'sig'                                                                                        |
| countType          | Default 'cooccur'. Can be 'all', 'cooccur', 'mutexcl'                                                                       |
| countsFontSize     | Default 0.8                                                                                                                 |
| countsFontColor    | Default 'black'                                                                                                             |
| colPal             | colPalBrewer palettes. See <a href="#">RColorBrewer::display.brewer.all()</a> for details                                   |
| showSum            | show [sum] with gene names in plot, Default TRUE                                                                            |
| plotPadj           | Plot adj. p-values instead                                                                                                  |
| colNC              | Number of different colors in the palette, minimum 3, default 9                                                             |
| nShiftSymbols      | shift if positive shift SigSymbols by n to the left, default = 5                                                            |
| sigSymbolsSize     | size of symbols in the matrix and in legend                                                                                 |
| sigSymbolsFontSize | size of font in legends                                                                                                     |
| pvSymbols          | vector of pch numbers for symbols of p-value for upper and lower thresholds<br>c(upper, lower)                              |
| limitColorBreaks   | limit color to extreme values. Default TRUE                                                                                 |

## Details

This function and plotting is inspired from genetic interaction analysis performed in the published study combining gene expression and mutation data in MDS. See reference for details.

Value

list of data.tables

References

Gerstung M, Pellagatti A, Malcovati L, et al. Combining gene mutation with gene expression data improves outcome prediction in myelodysplastic syndromes. Nature Communications. 2015;6:5901. doi:10.1038/ncomms6901.

Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
somaticInteractions(maf = laml, top = 5)
```

---

|           |                           |
|-----------|---------------------------|
| subsetMaf | <i>Subset MAF objects</i> |
|-----------|---------------------------|

---

Description

Subsets MAF based on given conditions.

Usage

```
subsetMaf(
  maf,
  tsb = NULL,
  genes = NULL,
  query = NULL,
  clinQuery = NULL,
  ranges = NULL,
  mult = "first",
  fields = NULL,
  mafObj = TRUE,
  includeSyn = TRUE,
  isTCGA = FALSE,
  dropLevels = TRUE,
  restrictTo = "all"
)
```

Arguments

- maf                    an MAF object generated by [read.maf](#)
- tsb                    subset by these samples (Tumor Sample Barcodes)
- genes                  subset by these genes
- query                  query string. e.g, "Variant\_Classification == 'Missense\_Mutation'" returns only Missense variants.

|            |                                                                                                                                                                                                                                           |
|------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| clinQuery  | query by clinical variable.                                                                                                                                                                                                               |
| ranges     | subset by ranges. data.frame with 3 column (chr, start, end). Overlaps are identified by <a href="#">foverlaps</a> function with arguments 'type = within', 'mult = all', 'no-match = NULL'                                               |
| mult       | When multiple loci in 'ranges' match to the variants maf, mult=. controls which values are returned - "all" , "first" (default) or "last". This value is passed to 'mult' argument of <a href="#">foverlaps</a>                           |
| fields     | include only these fields along with necessary fields in the output                                                                                                                                                                       |
| mafObj     | returns output as MAF class <a href="#">MAF-class</a> . Default TRUE                                                                                                                                                                      |
| includeSyn | Default TRUE, only applicable when mafObj = FALSE. If mafObj = TRUE, synonymous variants will be stored in a seperate slot of MAF object.                                                                                                 |
| isTCGA     | Is input MAF file from TCGA source.                                                                                                                                                                                                       |
| dropLevels | Default TRUE.                                                                                                                                                                                                                             |
| restrictTo | restrict subset operations to these. Can be 'all', 'cnv', or 'mutations'. Default 'all'. If 'cnv' or 'mutations', subset operations will only be applied on copy-number or mutation data respectively, while retaining other parts as is. |

## Value

subset table or an object of class [MAF-class](#)

## See Also

[getFields](#)

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
##Select all Splice_Site mutations from DNMT3A and NPM1
subsetMaf(maf = laml, genes = c('DNMT3A', 'NPM1'),
query = "Variant_Classification == 'Splice_Site'")
##Select all variants with VAF above 30%
subsetMaf(maf = laml, query = "i_TumorVAF_WU > 30")
##Extract data for samples 'TCGA.AB.3009' and 'TCGA.AB.2933' but only include vaf filed.
subsetMaf(maf = laml, tsb = c('TCGA-AB-3009', 'TCGA-AB-2933'), fields = 'i_TumorVAF_WU')
##Subset by ranges
ranges = data.frame(chr = c("2", "17"), start = c(25457000, 7571720), end = c(25458000, 7590868))
subsetMaf(laml, ranges = ranges)
```

survGroup

*Predict genesets associated with survival***Description**

Predict genesets associated with survival

**Usage**

```
survGroup(
  maf,
  top = 20,
  genes = NULL,
  geneSetSize = 2,
  minSamples = 5,
  clinicalData = NULL,
  time = "Time",
  Status = "Status",
  verbose = TRUE,
  plot = FALSE
)
```

**Arguments**

|              |                                                                                                                             |
|--------------|-----------------------------------------------------------------------------------------------------------------------------|
| maf          | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                         |
| top          | If genes is NULL by default used top 20 genes                                                                               |
| genes        | Manual set of genes                                                                                                         |
| geneSetSize  | Default 2                                                                                                                   |
| minSamples   | minimum number of samples to be mutated to be considered for analysis. Default 5                                            |
| clinicalData | dataframe containing events and time to events. Default looks for clinical data in annotation slot of <a href="#">MAF</a> . |
| time         | column name containing time in clinicalData                                                                                 |
| Status       | column name containing status of patients in clinicalData. must be logical or numeric. e.g, TRUE or FALSE, 1 or 0.          |
| verbose      | Default TRUE                                                                                                                |
| plot         | Default FALSE If TRUE, generate KM plots of the genesets combinations.                                                      |

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml.clin <- system.file("extdata", "tcga_laml_annot.tsv", package = "maftools")
laml <- read.maf(maf = laml.maf, clinicalData = laml.clin)
survGroup(maf = laml, top = 20, geneSetSize = 1, time = "days_to_last_followup", Status = "Overall_Survival_Status")
```

---

|               |                                       |
|---------------|---------------------------------------|
| tcgaAvailable | <i>Prints available TCGA datasets</i> |
|---------------|---------------------------------------|

---

**Description**

Prints available TCGA cohorts

**Usage**

```
tcgaAvailable(repo = c("github", "gitee"))
```

**Arguments**

repo                      can be "github" (default) or "gitee". If 'github' fails to fetch, switch to 'gitee'

**See Also**

[tcgaLoad](#)

**Examples**

```
tcgaAvailable()
```

---

|             |                                                   |
|-------------|---------------------------------------------------|
| tcgaCompare | <i>Compare mutation load against TCGA cohorts</i> |
|-------------|---------------------------------------------------|

---

**Description**

Compares mutation load in input MAF against all of 33 TCGA cohorts derived from MC3 project.

**Usage**

```
tcgaCompare(
  maf,
  capture_size = NULL,
  tcga_capture_size = 35.8,
  cohortName = NULL,
  tcga_cohorts = NULL,
  primarySite = FALSE,
  col = c("gray70", "black"),
  bg_col = c("#EDF8B1", "#2C7FB8"),
  medianCol = "red",
  decreasing = FALSE,
  logscale = TRUE,
  rm_hyper = FALSE,
  rm_zero = TRUE,
```

```

    cohortFontSize = 0.8,
    axisFontSize = 0.8
  )

```

## Arguments

|                   |                                                                                                                                                   |
|-------------------|---------------------------------------------------------------------------------------------------------------------------------------------------|
| maf               | MAF object(s) generated by <code>read.maf</code>                                                                                                  |
| capture_size      | capture size for input MAF in MBs. Default NULL. If provided plot will be scaled to mutations per mb. TCGA capture size is assumed to be 35.8 mb. |
| tcga_capture_size | capture size for TCGA cohort in MB. Default 35.8. Do NOT change. See details for more information.                                                |
| cohortName        | name for the input MAF cohort. Default "Input"                                                                                                    |
| tcga_cohorts      | restrict tcga data to these cohorts.                                                                                                              |
| primarySite       | If TRUE uses primary site of cancer as labels instead of TCGA project IDs. Default FALSE.                                                         |
| col               | color vector for length 2 TCGA cohorts and input MAF cohort. Default gray70 and black.                                                            |
| bg_col            | background color. Default '#EDF8B1', '#2C7FB8'                                                                                                    |
| medianCol         | color for median line. Default red.                                                                                                               |
| decreasing        | Default FALSE. Cohorts are arranged in increasing mutation burden.                                                                                |
| logscale          | Default TRUE                                                                                                                                      |
| rm_hyper          | Remove hyper mutated samples (outliers)? Default FALSE                                                                                            |
| rm_zero           | Remove samples with zero mutations? Default TRUE                                                                                                  |
| cohortFontSize    | Default 0.8                                                                                                                                       |
| axisFontSize      | Default 0.8                                                                                                                                       |

## Details

Tumor mutation burden for TCGA cohorts is obtained from TCGA MC3 study. For consistency TMB is estimated by restricting variants within Agilent Sureselect capture kit of size 35.8 MB.

## Value

data.table with median mutations per cohort

## Source

TCGA MC3 file was obtained from <https://api.gdc.cancer.gov/data/1c8cfe5f-e52d-41ba-94da-f15ea1337efc>. See TCGAmutations R package for more details. Further downstream script to estimate TMB for each sample can be found in 'inst/scripts/estimate\_tcga\_tmb.R'

## References

Scalable Open Science Approach for Mutation Calling of Tumor Exomes Using Multiple Genomic Pipelines Kyle Ellrott, Matthew H. Bailey, Gordon Saksena, et. al. Cell Syst. 2018 Mar 28; 6(3): 271–281.e7. <https://doi.org/10.1016/j.cels.2018.03.002>

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
tcgaCompare(maf = laml, cohortName = "AML")
```

---

tcgaDriverBP

*Compare genes to known TCGA drivers and their biological pathways*

---

## Description

A small function which uses known cancer driver genes and their associated pathways from TCGA cohorts. See reference for details

## Usage

```
tcgaDriverBP(m, genes = NULL, top = 20, fontSize = 0.7)
```

## Arguments

|          |                                                                                  |
|----------|----------------------------------------------------------------------------------|
| m        | an <a href="#">MAF</a> object                                                    |
| genes    | genes to compare. Default 'NULL'.                                                |
| top      | Top number of genes to use. Mutually exclusive with 'genes' argument. Default 20 |
| fontSize | Default 0.7                                                                      |

## References

Bailey MH, Tokheim C, Porta-Pardo E, et al. Comprehensive Characterization of Cancer Driver Genes and Mutations . Cell. 2018;173(2):371–385.e18. doi:10.1016/j.cell.2018.02.060

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
tcgaDriverBP(m = laml)
```

---

|          |                            |
|----------|----------------------------|
| tcgaLoad | <i>Loads a TCGA cohort</i> |
|----------|----------------------------|

---

## Description

Loads the user mentioned TCGA cohorts

## Usage

```
tcgaLoad(  
  study = NULL,  
  source = c("MC3", "Firehose"),  
  repo = c("github", "gitee")  
)
```

## Arguments

|        |                                                                                                 |
|--------|-------------------------------------------------------------------------------------------------|
| study  | Study names to load. Use <a href="#">tcgaAvailable</a> to see available options.                |
| source | Source for MAF files. Can be MC3 or Firehose. Default MC3. Argument may be abbreviated (M or F) |
| repo   | one of "github" (default) and "gitee".                                                          |

## Details

The function loads curated and pre-compiled MAF objects from TCGA cohorts. TCGA data are obtained from two sources namely, Broad Firehose repository, and MC3 project.

## Value

An object of class MAF.

## References

Scalable Open Science Approach for Mutation Calling of Tumor Exomes Using Multiple Genomic Pipelines Kyle Ellrott, Matthew H. Bailey, Gordon Saksena, et. al. Cell Syst. 2018 Mar 28; 6(3): 271–281.e7.

## See Also

[tcgaAvailable](#)

## Examples

```
# Loads TCGA LAML cohort (default from MC3 project)  
tcgaLoad(study = "LAML")  
# Loads TCGA LAML cohort (from Broad Firehose)  
tcgaLoad(study = "LAML", source = "Firehose")
```

---

|      |                                                           |
|------|-----------------------------------------------------------|
| titv | <i>Classifies SNPs into transitions and transversions</i> |
|------|-----------------------------------------------------------|

---

## Description

takes output generated by `read.maf` and classifies Single Nucleotide Variants into Transitions and Transversions.

## Usage

```
titv(maf, useSyn = FALSE, plot = TRUE, file = NULL)
```

## Arguments

|        |                                                                                        |
|--------|----------------------------------------------------------------------------------------|
| maf    | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                    |
| useSyn | Logical. Whether to include synonymous variants in analysis. Defaults to FALSE.        |
| plot   | plots a titv fractions. default TRUE.                                                  |
| file   | basename for output file name. If given writes summaries to output file. Default NULL. |

## Value

list of data.frames with Transitions and Transversions summary.

## See Also

[plotTiTv](#)

## Examples

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.titv = titv(maf = laml, useSyn = TRUE)
```

---

|     |                                       |
|-----|---------------------------------------|
| tmb | <i>Estimate Tumor Mutation Burden</i> |
|-----|---------------------------------------|

---

**Description**

Estimates Tumor Mutation Burden in terms of per megabases

**Usage**

```
tmb(maf, captureSize = 50, logScale = TRUE)
```

**Arguments**

|             |                                                  |
|-------------|--------------------------------------------------|
| maf         | maf <a href="#">MAF</a> object                   |
| captureSize | capture size for input MAF in MBs. Default 50MB. |
| logScale    | Default TRUE. For plotting purpose only.         |

**Value**

data.table with TMB for every sample

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
tmb(maf = laml)
```

---

|                     |                                                                                                                                          |
|---------------------|------------------------------------------------------------------------------------------------------------------------------------------|
| trinucleotideMatrix | <i>Extract single 5' and 3' bases flanking the mutated site for de-novo signature analysis. Also estimates APOBEC enrichment scores.</i> |
|---------------------|------------------------------------------------------------------------------------------------------------------------------------------|

---

**Description**

Extract single 5' and 3' bases flanking the mutated site for de-novo signature analysis. Also estimates APOBEC enrichment scores.

**Usage**

```
trinucleotideMatrix(
  maf,
  ref_genome = NULL,
  prefix = NULL,
  add = TRUE,
  ignoreChr = NULL,
  useSyn = TRUE,
  fn = NULL
)
```

**Arguments**

|            |                                                                                                                                                             |
|------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|
| maf        | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a>                                                                                         |
| ref_genome | BSgenome object or name of the installed BSgenome package. Example: BSgenome.Hsapiens.UCSC.hg19. Default NULL, tries to auto-detect from installed genomes. |
| prefix     | Prefix to add or remove from contig names in MAF file.                                                                                                      |
| add        | If prefix is used, default is to add prefix to contig names in MAF file. If false prefix will be removed from contig names.                                 |
| ignoreChr  | Chromosomes to ignore from analysis. e.g. chrM                                                                                                              |
| useSyn     | Logical. Whether to include synonymous variants in analysis. Defaults to TRUE                                                                               |
| fn         | If given writes APOBEC results to an output file with basename fn. Default NULL.                                                                            |

**Details**

Extracts immediate 5' and 3' bases flanking the mutated site and classifies them into 96 substitution classes. Requires BSgenome data packages for sequence extraction.

APOBEC Enrichment: Enrichment score is calculated using the same method described by Roberts et al.

$$E = (n\_tcw * background\_c) / (n\_C * background\_tcw)$$

where, n\_tcw = number of mutations within T[C>T]W and T[C>G]W context. (W -> A or T)

n\_C = number of mutated C and G

background\_C and background\_tcw motifs are number of C and TCW motifs occurring around +/- 20bp of each mutation.

One-sided Fisher's Exact test is performed to determine the enrichment of APOBEC tcw mutations over background.

**Value**

list of 2. A matrix of dimension nx96, where n is the number of samples in the MAF and a table describing APOBEC enrichment per sample.

**References**

Roberts SA, Lawrence MS, Klimczak LJ, et al. An APOBEC Cytidine Deaminase Mutagenesis Pattern is Widespread in Human Cancers. Nature genetics. 2013;45(9):970-976. doi:10.1038/ng.2702.

**See Also**

[extractSignatures](#) [plotApobecDiff](#)

## Examples

```
## Not run:
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
laml.tnm <- trinucleotideMatrix(maf = laml, ref_genome = 'BSgenome.Hsapiens.UCSC.hg19',
prefix = 'chr', add = TRUE, useSyn = TRUE)

## End(Not run)
```

---

|            |                                       |
|------------|---------------------------------------|
| vafCompare | <i>compare VAF across two cohorts</i> |
|------------|---------------------------------------|

---

## Description

Draw boxplot distribution of VAFs across two cohorts

## Usage

```
vafCompare(
  m1,
  m2,
  genes = NULL,
  top = 5,
  vafCol1 = NULL,
  vafCol2 = NULL,
  m1Name = "M1",
  m2Name = "M2",
  cols = c("#2196F3", "#4CAF50"),
  sigvals = TRUE,
  nrows = NULL,
  ncols = NULL
)
```

## Arguments

|         |                                                                               |
|---------|-------------------------------------------------------------------------------|
| m1      | first <a href="#">MAF</a> object. Required.                                   |
| m2      | second <a href="#">MAF</a> object. Required.                                  |
| genes   | specify genes for which plot has to be generated. Default NULL.               |
| top     | if genes is NULL plots top n number of genes. Defaults to 5.                  |
| vafCol1 | manually specify column name for vafs in m1. Default looks for column 't_vaf' |
| vafCol2 | manually specify column name for vafs in m2. Default looks for column 't_vaf' |
| m1Name  | optional name for first cohort                                                |
| m2Name  | optional name for second cohort                                               |
| cols    | vector of colors corresponding to m1 and m2 respectively.                     |

|         |                                                                      |
|---------|----------------------------------------------------------------------|
| sigvals | Estimate and add significance stars. Default TRUE.                   |
| nrows   | Number of rows in the layout. Default NULL - estimated automatically |
| ncols   | Number of genes drawn per row. Default 4                             |

---

|                     |                                                                    |
|---------------------|--------------------------------------------------------------------|
| write.GisticSummary | <i>Writes GISTIC summaries to output tab-delimited text files.</i> |
|---------------------|--------------------------------------------------------------------|

---

## Description

Writes GISTIC summaries to output tab-delimited text files.

## Usage

```
write.GisticSummary(gistic, basename = NULL)
```

## Arguments

|          |                                                   |
|----------|---------------------------------------------------|
| gistic   | an object of class GISTIC generated by readGistic |
| basename | basename for output file to be written.           |

## Value

None. Writes output as tab delimited text files.

## See Also

[readGistic](#)

## Examples

```
all.lesions <- system.file("extdata", "all_lesions.conf_99.txt", package = "maftools")
amp.genes <- system.file("extdata", "amp_genes.conf_99.txt", package = "maftools")
del.genes <- system.file("extdata", "del_genes.conf_99.txt", package = "maftools")
scores.gistic <- system.file("extdata", "scores.gistic", package = "maftools")
laml.gistic = readGistic(gisticAllLesionsFile = all.lesions, gisticAmpGenesFile = amp.genes, gisticDelGenesFile =
write.GisticSummary(gistic = laml.gistic, basename = 'laml')
```

---

|                  |                                                                 |
|------------------|-----------------------------------------------------------------|
| write.mafSummary | <i>Writes maf summaries to output tab-delimited text files.</i> |
|------------------|-----------------------------------------------------------------|

---

**Description**

Writes maf summaries to output tab-delimited text files.

**Usage**

```
write.mafSummary(maf, basename = NULL, compress = FALSE)
```

**Arguments**

|          |                                                                     |
|----------|---------------------------------------------------------------------|
| maf      | an <a href="#">MAF</a> object generated by <a href="#">read.maf</a> |
| basename | basename for output file to be written.                             |
| compress | If 'TRUE' files will be gz compressed. Default 'FALSE'              |

**Details**

Writes MAF and related summaries to output files.

**Value**

None. Writes output as text files.

**See Also**

[read.maf](#)

**Examples**

```
laml.maf <- system.file("extdata", "tcga_laml.maf.gz", package = "maftools")
laml <- read.maf(maf = laml.maf)
write.mafSummary(maf = laml, basename = 'laml')
```

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