

Package ‘CaMutQC’

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Type Package

Title An R Package for Comprehensive Filtration and Selection of
Cancer Somatic Mutations

Version 1.1.1

Description CaMutQC is able to filter false positive mutations generated due to technical issues, as well as to select candidate cancer mutations through a series of well-structured functions by labeling mutations with various flags. And a detailed and vivid filter report will be offered after completing a whole filtration or selection section. Also, CaMutQC integrates several methods and gene panels for Tumor Mutational Burden (TMB) estimation.

biocViews Software, QualityControl, GeneTarget

Encoding UTF-8

LazyData false

Suggests knitr, rmarkdown, BiocStyle

VignetteBuilder knitr

RoxygenNote 7.3.1

NeedsCompilation no

BugReports <https://github.com/likelet/CaMutQC/issues>

URL <https://github.com/likelet/CaMutQC>

Depends R (>= 4.0.0)

Imports ggplot2, dplyr, org.Hs.eg.db, vcfR, clusterProfiler, stringr,
DT, MesKit, maftools, data.table, utils, stats, methods, tidy

License GPL-3

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Author Xin Wang [aut, cre] (<<https://orcid.org/0000-0002-6072-599X>>)

Maintainer Xin Wang <sylviawang555@gmail.com>

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calTMB

calTMB

Description

Calculate Tumor Mutational Burden (TMB) in specific regions.

Usage

```
calTMB(
  maf,
  bedFile = NULL,
  bedHeader = FALSE,
  assay = "MSK-v3",
  genelist = NULL,
  mutType = "nonsynonymous",
  bedFilter = TRUE
)
```

Arguments

| | |
|-----------|--|
| maf | An MAF data frame, generated by vcfToMAF function. |
| bedFile | A file in bed format that contains region information. Default: NULL. |
| bedHeader | Whether the input bed file has a header or not. Default: FALSE. |
| assay | Methodology and assay will be applied as a reference, including 'MSK-v3', 'MSK-v2', 'MSK-v1', 'FoundationOne', 'Pan-Cancer Panel' and 'Customized'. Default: 'MSK-v3'. |
| genelist | A vector of panel gene list, only useful when assay is set to 'Customized'. |

| | |
|-----------|---|
| mutType | A group of variant classifications that will be kept, only useful when assay is set to 'Pan-Cancer Panel' or 'Customized', including 'exonic', 'nonsynonymous', and 'all' Default: 'nonsynonymous'. |
| bedFilter | Whether to filter the information in bed file or not, which only leaves segments in Chr1-Ch22, ChrX and ChrY. Default: TRUE. |

Value

A TMB value.

Examples

```
maf <- vcfToMAF(system.file("extdata", "WES_EA_T_1_mutect2.vcf",
package="CaMutQC"))
TMB_value <- calTMB(maf, bedFile=system.file("extdata/bed/panel_hg38",
"F1CDx-hg38.rds", package="CaMutQC"))
```

| | |
|--------------|---------------------|
| mutFilterAdj | <i>mutFilterAdj</i> |
|--------------|---------------------|

Description

Filter SNVs with adjacent indels

Usage

```
mutFilterAdj(maf, maxIndelLen = 50, minInterval = 10)
```

Arguments

| | |
|-------------|---|
| maf | An MAF data frame, generated by vcfToMAF function. |
| maxIndelLen | Maximum length of indel accepted to be included. Default: 50 |
| minInterval | Minimum length of interval between an SNV and an indel accepted to be included. Default: 10 |

Value

An MAF data frame after filtration for adjacent variants.

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
mafF <- mutFilterAdj(maf)
```

| | |
|--------------|---------------------|
| mutFilterCan | <i>mutFilterCan</i> |
|--------------|---------------------|

Description

Apply common filtering strategies on a MAF data frame for different cancer types.

Usage

```
mutFilterCan(  
  maf,  
  cancerType,  
  PONfile,  
  PONformat = "vcf",  
  panel = "Customized",  
  tumorDP = 0,  
  normalDP = 0,  
  tumorAD = 0,  
  normalAD = Inf,  
  VAF = 0,  
  VAFratio = 0,  
  SBmethod = "SOR",  
  SBscore = Inf,  
  maxIndelLen = Inf,  
  minInterval = 0,  
  tagFILTER = NULL,  
  dbVAF = 0.01,  
  ExAC = FALSE,  
  Genomesprojects1000 = FALSE,  
  ESP6500 = FALSE,  
  gnomAD = FALSE,  
  dbSNP = FALSE,  
  keepCOSMIC = FALSE,  
  keepType = "all",  
  bedFile = NULL,  
  bedFilter = TRUE,  
  bedHeader = FALSE,  
  mutFilter = FALSE,  
  selectCols = FALSE,  
  report = TRUE,  
  reportFile = "FilterReport.html",  
  reportDir = "./",  
  TMB = FALSE,  
  progressbar = TRUE,  
  codelog = FALSE,  
  codelogFile = "mutFilterCan.log",  
  verbose = TRUE  
)
```

Arguments

`maf` An MAF data frame.

| | |
|---------------------|--|
| cancerType | Type of cancer whose filtering parameters need to be referred to. Options are: "COADREAD", "BRCA", "LIHC", "LAML", "LCML", "UCEC", "UCS", "BLCA", "KIRC" and "KIRP" |
| PONfile | Panel-of-Normals files, which can be either obtained through GATK (https://gatk.broadinstitute.org/hc/en-us/articles/360035890631-Panel-of-Normals-PON-) or generated by users. Should have at least four columns: CHROM, POS, REF, ALT |
| PONformat | The format of PON file, either "vcf" or "txt". Default: "vcf" |
| panel | The sequencing panel applied on the dataset. Parameters for <code>mutFilterQual</code> function are set differently for different panels. Default: "Customized". Options: "MSKCC", "WES". |
| tumorDP | Threshold of tumor total depth. Default: 20 |
| normalDP | Threshold of normal total depth. Default: 10 |
| tumorAD | Threshold of tumor alternative allele depth. Default:5 |
| normalAD | Threshold of normal alternative allele depth. Default: Inf |
| VAF | Threshold of VAF value. Default: 0.05 |
| VAFratio | Threshold of VAF ratio (tVAF/nVAF). Default: 0 |
| SBmethod | Method will be used to detect strand bias, including 'SOR' and 'Fisher'. Default: 'SOR'. SOR: StrandOddsRatio (https://gatk.broadinstitute.org/hc/en-us/articles/360041849111-StrandOddsRatio) |
| SBscore | Cutoff strand bias score used to filter variants. Default: 3 |
| maxIndelLen | Maximum length of indel accepted to be included. Default: 50 |
| minInterval | Maximum length of interval between an SNV and an indel accepted to be included. Default: 10 |
| tagFILTER | Variants with specific tag in the FILTER column will be kept, Default: 'PASS' |
| dbVAF | Threshold of VAF of certain population for variants in database. Default: 0.01. |
| ExAC | Whether to filter variants listed in ExAC with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| Genomesprojects1000 | Whether to filter variants listed in Genomesprojects1000 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| ESP6500 | Whether to filter variants listed in ESP6500 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| gnomAD | Whether to filter variants listed in gnomAD with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| dbSNP | Whether to filter variants listed in dbSNP. Default: FALSE. |
| keepCOSMIC | Whether to keep variants in COSMIC even they have are present in germline database. Default: TRUE. |
| keepType | A group of variant classifications will be kept, including 'exonic', 'nonsynonymous' and 'all'. Default: 'exonic'. |
| bedFile | A file in bed format that contains region information. Default: NULL |
| bedFilter | Whether to filter the information in bed file or not, which only leaves segments in Chr1-Ch22, ChrX and ChrY. Default: TRUE |
| bedHeader | Whether the input bed file has a header or not. Default: FALSE. |

| | |
|-------------|--|
| mutFilter | Whether to directly return a filtered MAF data frame. If FALSE, a simulation filtration process will be run, and the original MAF data frame with tags in CaTag column, and a filter report will be returned. If TRUE, a filtered MAF data frame and a filter report will be generated. Default: FALSE |
| selectCols | Columns will be contained in the filtered data frame. By default (TRUE), the first 13 columns and 'Tumor_Sample_Barcode' column. Or a vector contains column names will be kept. |
| report | Whether to generate report automatically. Default: TRUE |
| reportFile | File name of the report. Default: 'FilterReport.html' |
| reportDir | Path to the output report file. Default: './' |
| TMB | Whether to calculate TMB. Default: TRUE |
| progressbar | Whether to show progress bar when running this function Default: TRUE |
| codelog | If TRUE, your code, along with the parameters you set, will be export in a log file. It will be convenient for users to repeat experiments. Default: FALSE |
| codelogFile | Where to store the codelog, only useful when codelog is set to TRUE. Default: "mutFilterCan.log" |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame after common strategy filtration for a cancer type.

A filter report in HTML format

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
mafF <- mutFilterCan(maf, cancerType='BRCA',
PONfile=system.file("extdata", "PON_test.txt", package="CaMutQC"),
PONformat="txt", TMB=FALSE)
```

mutFilterCom

mutFilterCom

Description

Apply common filtering strategies on a MAF data frame.

Usage

```
mutFilterCom(
  maf,
  PONfile,
  PONformat = "vcf",
  panel = "Customized",
  tumorDP = 20,
  normalDP = 10,
  tumorAD = 5,
```

```

normalAD = Inf,
VAF = 0.05,
VAFratio = 0,
SBmethod = "SOR",
SBscore = 3,
maxIndelLen = 50,
minInterval = 10,
tagFILTER = "PASS",
dbVAF = 0.01,
ExAC = TRUE,
Genomesprojects1000 = TRUE,
gnomAD = TRUE,
dbSNP = FALSE,
keepCOSMIC = TRUE,
keepType = "exonic",
bedFile = NULL,
bedHeader = FALSE,
bedFilter = TRUE,
mutFilter = FALSE,
ESP6500 = TRUE,
selectCols = TRUE,
report = TRUE,
assay = "MSK-v3",
genelist = NULL,
mutType = "nonsynonymous",
reportFile = "FilterReport.html",
reportDir = "./",
TMB = TRUE,
cancerType = NULL,
reference = NULL,
progressbar = TRUE,
codelog = FALSE,
codelogFile = "mutFilterCom.log",
verbose = TRUE
)

```

Arguments

| | |
|-----------|--|
| maf | An MAF data frame. |
| PONfile | Panel-of-Normals files, which can be either obtained through GATK (https://gatk.broadinstitute.org/hc/en-us/articles/360035890631-Panel-of-Normals-PON-) or generated by users. Should have at least four columns: CHROM, POS, REF, ALT |
| PONformat | The format of PON file, either "vcf" or "txt". Default: "vcf" |
| panel | The sequencing panel applied on the dataset. Parameters for <code>mutFilterQual</code> function are set differently for different panels. Default: "Customized". Options: "MSKCC", "WES". |
| tumorDP | Threshold of tumor total depth. Default: 20 |
| normalDP | Threshold of normal total depth. Default: 10 |
| tumorAD | Threshold of tumor alternative allele depth. Default: 5 |
| normalAD | Threshold of normal alternative allele depth. Default: Inf |

| | |
|---------------------|---|
| VAF | Threshold of VAF value. Default: 0.05 |
| VAFRatio | Threshold of VAF ratio (tVAF/nVAF). Default: 0. |
| SBmethod | Method will be used to detect strand bias, including 'SOR' and 'Fisher'. Default: 'SOR'. SOR: StrandOddsRatio (https://gatk.broadinstitute.org/hc/en-us/articles/360041849111 StrandOddsRatio) |
| SBscore | Cutoff strand bias score used to filter variants. Default: 3. |
| maxIndelLen | Maximum length of indel accepted to be included. Default: 50. |
| minInterval | Maximum length of interval between an SNV and an indel accepted to be included. Default: 10. |
| tagFILTER | Variants with specific tag in the FILTER column will be kept, Default: 'PASS'. |
| dbVAF | Threshold of VAF value for databases. Default: 0.01. |
| ExAC | Whether to filter variants listed in ExAC with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| Genomesprojects1000 | Whether to filter variants listed in Genomesprojects1000 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| gnomAD | Whether to filter variants listed in gnomAD with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| dbSNP | Whether to filter variants listed in dbSNP. Default: FALSE. |
| keepCOSMIC | Whether to keep variants in COSMIC even they have are present in germline database. Default: TRUE. |
| keepType | A group of variant classifications will be kept, including 'exonic', 'nonsynonymous' and 'all'. Default: 'exonic'. |
| bedFile | A file in bed format that contains region information. Default: NULL. |
| bedHeader | Whether the input bed file has a header or not. Default: FALSE. |
| bedFilter | Whether to filter the information in bed file or not, which only leaves segments in Chr1-Ch22, ChrX and ChrY. Default: TRUE. |
| mutFilter | Whether to directly return a filtered MAF data frame. If FALSE, a simulation filtration process will be run, and the original MAF data frame with tags in CaTag column, and a filter report will be returned. If TRUE, a filtered MAF data frame and a filter report will be generated. Default: FALSE. |
| ESP6500 | Whether to filter variants listed in ESP6500 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| selectCols | Columns will be contained in the filtered data frame. By default (TRUE), the first 13 columns and 'Tumor_Sample_Barcode' column. Or a vector contains column names will be kept. |
| report | Whether to generate report automatically. Default: TRUE |
| assay | Methodology and assay will be applied as a reference, including 'MSK-v3', 'MSK-v2', 'MSK-v1', 'FoundationOne', 'Pan-Cancer Panel' and 'Customized'. Default: 'MSK-v3'. |
| genelist | A vector of panel gene list, only useful when assay is set to 'Customized'. |
| mutType | A group of variant classifications that will be kept, only useful when assay is set to 'Pan-Cancer Panel' or 'Customized', including 'exonic' and 'nonsynonymous'. Default: 'nonsynonymous'. |
| reportFile | File name of the report. Default: 'FilterReport.html' |

| | |
|-------------|--|
| reportDir | Path to the output report file. Default: './'. |
| TMB | Whether to calculate TMB. Default: TRUE. Note: CaMutQC uses unfiltered maf to calculate TMB value. |
| cancerType | Type of cancer whose filtering parameters need to be referred to. Options are: "COADREAD", "BRCA", "LIHC", "LAML", "LCML", "UCEC", "UCS", "BLCA", "KIRC" and "KIRP" |
| reference | A specific study whose filtering strategies need to be referred to. Format: "Last_name_of_the_first_author-Journal-Year-Cancer_type" Options are: "Haraltdottir_et_al-Gastroenterology-2014-UCEC", "Cherniack_et_al-Cancer_Cell-2017-UCS", "Mason_et_al-Leukemia-2015-LCML", "Gerlinger_et_al-Engl_J_Med-2012-KIRC" "Zhu_et_al-Nat_Communications-2020-KIRP" |
| progressbar | Whether to show progress bar when running this function Default: TRUE |
| codelog | If TRUE, your code, along with the parameters you set, will be export in a log file. It will be convenient for users to repeat experiments. Default: FALSE |
| codelogFile | Where to store the codelog, only useful when codelog is set to TRUE. Default: "mutFilterCom.log" |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame after common strategy filtration

A filter report in HTML format

Examples

```
maf <- vcfToMAF(system.file("extdata",
  "WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
maff <- mutFilterCom(maf,
  PONfile=system.file("extdata", "PON_test.txt", package="CaMutQC"),
  TMB=FALSE, report=FALSE, PONformat="txt", verbose=FALSE)
```

mutFilterDB

mutFilterDB

Description

Filter variants in germline database.

Usage

```
mutFilterDB(
  maf,
  dbVAF = 0.01,
  ExAC = TRUE,
  Genomesprojects1000 = TRUE,
  ESP6500 = TRUE,
  gnomAD = TRUE,
  dbSNP = FALSE,
  keepCOSMIC = TRUE,
  verbose = TRUE
)
```

Arguments

| | |
|---------------------|---|
| maf | An MAF data frame, generated by <code>vcfToMAF</code> function. |
| dbVAF | Threshold of VAF value for database annotations. Default: 0.01. |
| ExAC | Whether to filter variants listed in ExAC with VAF higher than cutoff (set in dbVAF parameter). Default: TRUE. |
| Genomesprojects1000 | Whether to filter variants listed in Genomesprojects1000 with VAF higher than cutoff (set in dbVAF parameter). Default: TRUE. |
| ESP6500 | Whether to filter variants listed in ESP6500 with VAF higher than cutoff (set in dbVAF parameter). Default: TRUE. |
| gnomAD | Whether to filter variants listed in gnomAD with VAF higher than cutoff (set in dbVAF parameter). Default: TRUE. |
| dbSNP | Whether to filter variants listed in dbSNP. Default: FALSE. |
| keepCOSMIC | Whether to keep variants in COSMIC even they are present in germline database. Default: TRUE. |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame after filtration for database and clinical significance

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
mafF <- mutFilterDB(maf)
```

mutFilterNormalDP *mutFilterNormalDP*

Description

Filter dbSNP/non-dbSNP variants based on their normal depth. Variants in dbSNP database should have normal depth ≥ 19 , while non-dbSNP variants should have normal depth ≥ 8 to avoid being filtered.

Usage

```
mutFilterNormalDP(maf, dbSNPCutoff = 19, nonCutoff = 8, verbose = TRUE)
```

Arguments

| | |
|-------------|--|
| maf | An MAF data frame, generated by <code>vcfToMAF</code> function. |
| dbSNPCutoff | Cutoff of normal depth for dbSNP variants. Default: 19. |
| nonCutoff | Cutoff of normal depth for non-dbSNP variants. Default: 8. |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame where some variants has N tag in CaTag column for Normal depth filtration.

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
maff <- mutFilterNormalDP(maf)
```

| | |
|--------------|---------------------|
| mutFilterPON | <i>mutFilterPON</i> |
|--------------|---------------------|

Description

Filter variants based on Panel of Normals

Usage

```
mutFilterPON(maf, PONfile, PONformat = "vcf", verbose = TRUE)
```

Arguments

| | |
|-----------|--|
| maf | An MAF data frame, generated by <code>vcfToMAF</code> function. |
| PONfile | Panel-of-Normals files, which can be either obtained through GATK (https://gatk.broadinstitute.org/hc/en-us/articles/360035890631-Panel-of-Normals-PON-) or generated by users. Should have at least four columns: CHROM, POS, REF, ALT |
| PONformat | The format of PON file, either "vcf" or "txt". Default: "vcf" |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame where some variants have P tag in CaTag column for PON filtration.

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
maff <- mutFilterPON(maf, PONfile=system.file("extdata",
"PON_test.txt", package="CaMutQC"), PONformat="txt")
```

mutFilterQual

mutFilterQual

Description

Filter variants in low sequencing quality or low confidence.

Usage

```
mutFilterQual(
  maf,
  panel = "Customized",
  tumorDP = 20,
  normalDP = 10,
  tumorAD = 5,
  normalAD = Inf,
  VAF = 0.05,
  VAFratio = 0
)
```

Arguments

| | |
|----------|--|
| maf | An MAF data frame, generated by vcfToMAF function. |
| panel | The sequencing panel applied on the dataset. Parameters for mutFilterQual function are set differently for different panels. Default: "Customized". Options: "MSKCC", "WES". |
| tumorDP | Threshold of tumor total depth. Default: 20 |
| normalDP | Threshold of normal total depth. Default: 10 |
| tumorAD | Threshold of tumor alternative allele depth. Default: 5 |
| normalAD | Threshold of normal alternative allele depth. Default: Inf |
| VAF | Threshold of VAF value. Default: 0.05 |
| VAFratio | Threshold of VAF ratio (tVAF/nVAF). Default: 0 |

Value

An MAF data frame where some variants have Q tag in CaTag column for sequencing quality filtration

Examples

```
maf <- vcfToMAF(system.file("extdata",
  "WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
mafF <- mutFilterQual(maf)
```

| | |
|--------------|---------------------|
| mutFilterRef | <i>mutFilterRef</i> |
|--------------|---------------------|

Description

Use the same filtering strategies that a specific study used, or top-rated strategies shared by users.

Usage

```
mutFilterRef(  
  maf,  
  reference,  
  PONfile,  
  PONformat = "vcf",  
  tumorDP = 0,  
  normalDP = 0,  
  tumorAD = 0,  
  normalAD = Inf,  
  VAF = 0,  
  VAFratio = 0,  
  SBmethod = "SOR",  
  SBscore = Inf,  
  maxIndelLen = Inf,  
  minInterval = 0,  
  tagFILTER = NULL,  
  dbVAF = 0.01,  
  ExAC = FALSE,  
  Genomesprojects1000 = FALSE,  
  ESP6500 = FALSE,  
  gnomAD = FALSE,  
  dbSNP = FALSE,  
  keepCOSMIC = FALSE,  
  keepType = "all",  
  bedFile = NULL,  
  bedFilter = TRUE,  
  mutFilter = FALSE,  
  selectCols = FALSE,  
  report = TRUE,  
  reportFile = "FilterReport.html",  
  reportDir = "./",  
  TMB = FALSE,  
  progressBar = TRUE,  
  codelog = FALSE,  
  codelogFile = "mutFilterCom.log",  
  verbose = TRUE  
)
```

Arguments

maf An MAF data frame.

| | |
|---------------------|--|
| reference | A specific study whose filtering strategies need to be referred to. Format: "Last_name_of_the_first_author-Journal-Year-Cancer_type" Options are: "Haraldsdottir_et_al-Gastroenterology-2014-UCEC", "Cherniack_et_al-Cancer_Cell-2017-UCS", "Mason_et_al-Leukemia-2015-LCML", "Gerlinger_et_al-Engl_J_Med-2012-KIRC", "Zhu_et_al-Nat_Comm-2020-KIRP" |
| PONfile | Panel-of-Normals files, which can be either obtained through GATK (https://gatk.broadinstitute.org/hc/en-us/articles/360035890631-Panel-of-Normals-PON-) or generated by users. Should have at least four columns: CHROM, POS, REF, ALT |
| PONformat | The format of PON file, either "vcf" or "txt". Default: "vcf" |
| tumorDP | Threshold of tumor total depth. Default: 0 |
| normalDP | Threshold of normal total depth. Default: 0 |
| tumorAD | Threshold of tumor alternative allele depth. Default: 0 |
| normalAD | Threshold of normal alternative allele depth. Default: Inf |
| VAF | Threshold of VAF value. Default: 0 |
| VAFRatio | Threshold of VAF ratio (tVAF/nVAF). Default: 0 |
| SBmethod | Method will be used to detect strand bias, including 'SOR' and 'Fisher'. Default: 'SOR'. SOR: StrandOddsRatio (https://gatk.broadinstitute.org/hc/en-us/articles/360041849111-StrandOddsRatio) |
| SBscore | Cutoff strand bias score used to filter variants. Default: 3 |
| maxIndelLen | Maximum length of indel accepted to be included. Default: Inf |
| minInterval | Maximum length of interval between an SNV and an indel accepted to be included. Default: 0 |
| tagFILTER | Variants with specific tag in the FILTER column will be kept, Default: NULL |
| dbVAF | Threshold of VAF of certain population for variants in database. Default: 0.01 |
| ExAC | Whether to filter variants listed in ExAC with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| Genomesprojects1000 | Whether to filter variants listed in Genomesprojects1000 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| ESP6500 | Whether to filter variants listed in ESP6500 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| gnomAD | Whether to filter variants listed in gnomAD with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| dbSNP | Whether to filter variants listed in dbSNP. Default: FALSE. |
| keepCOSMIC | Whether to keep variants in COSMIC even they have are present in germline database. Default: FALSE. |
| keepType | A group of variant classifications will be kept, including 'exonic', 'nonsynonymous' and 'all'. Default: 'all'. |
| bedFile | A file in bed format that contains region information. Default: NULL. |
| bedFilter | Whether to filter the information in bed file or not, which only leaves segments in Chr1-Ch22, ChrX and ChrY. Default: TRUE |
| mutFilter | Whether to directly return a filtered MAF data frame. If FALSE, a simulation filtration process will be run, and the original MAF data frame with tags in CaTag column, and a filter report will be returned. If TRUE, a filtered MAF data frame and a filter report will be generated. Default: FALSE |

| | |
|-------------|--|
| selectCols | Columns will be contained in the filtered data frame. By default (TRUE), the first 13 columns and 'Tumor_Sample_Barcode' column. Or a vector contains column names will be kept. |
| report | Whether to generate report automatically. Default: TRUE |
| reportFile | File name of the report. Default: 'FilterReport.html' |
| reportDir | Path to the output report file. Default: './' |
| TMB | Whether to calculate TMB. Default: TRUE |
| progressbar | Whether to show progress bar when running this function Default: TRUE |
| codelog | If TRUE, your code, along with the parameters you set, will be export in a log file. It will be convenient for users to repeat experiments. Default: FALSE |
| codelogFile | Where to store the codelog, only useful when codelog is set to TRUE. Default: "mutFilterCom.log" |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame after applied filtering strategies in another study

A filter report in HTML format

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf",package="CaMutQC"))
mafR <- mutFilterRef(maf, reference="Zhu_et_al-Nat_Commun-2020-KIRP",
PONfile=system.file("extdata","PON_test.txt", package="CaMutQC"),
PONformat="txt", TMB=FALSE, verbose=FALSE, report=FALSE)
```

mutFilterReg

mutFilterReg

Description

Filter variants not in specific regions.

Usage

```
mutFilterReg(
  maf,
  bedFile = NULL,
  bedHeader = FALSE,
  bedFilter = TRUE,
  verbose = TRUE
)
```

Arguments

| | |
|-----------|---|
| maf | An MAF data frame, generated by <code>vcfToMAF</code> function. |
| bedFile | A bed file that contains region information. Default: NULL |
| bedHeader | Whether the input bed file has a header or not. Default: FALSE. |
| bedFilter | Whether to filter the information in bed file or not, which only leaves segments in Chr1-Ch22, ChrX and ChrY. Default: TRUE |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame where some variants have R tag in CaTag column for region filtration.

Examples

```
maf <- vcfToMAF(system.file("extdata", "WES_EA_T_1_mutect2.vcf",
package="CaMutQC"))
mafF <- mutFilterReg(maf, bedFile=system.file("extdata/bed/panel_hg38",
"Pan-cancer-hg38.rds", package="CaMutQC"))
```

mutFilterSB

mutFilterSB

Description

Filter variants based on strand bias.

Usage

```
mutFilterSB(maf, method = "SOR", SBscore = 3)
```

Arguments

| | |
|---------|---|
| maf | An MAF object, generated by <code>vcfToMAF</code> function. |
| method | Method will be used to detect strand bias, including 'SOR' and 'Fisher'. Default: 'SOR'. SOR: StrandOddsRatio (https://gatk.broadinstitute.org/hc/en-us/articles/360041849111) Fisher's Exat Test: Switch to Phred socre (https://gatk.broadinstitute.org/hc/en-us/articles/360035532152-Fisher-s-Exact-Test) |
| SBscore | Cutoff strand bias score used to filter variants. Default: 3 |

Value

An MAF data frame where some variants have S tag in CaTag column for strand bias filtration

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
mafF <- mutFilterSB(maf)
```

| | |
|---------------|----------------------|
| mutFilterTech | <i>mutFilterTech</i> |
|---------------|----------------------|

Description

Filter potential artifacts produced through technical issue, including filtration for sequencing quality, strand bias, adjacent indel tag, normal depth, panel of normal (PON) and FILTER field.

Usage

```
mutFilterTech(
  maf,
  PONfile,
  PONformat = "vcf",
  panel = "Customized",
  tumorDP = 20,
  normalDP = 10,
  tumorAD = 5,
  normalAD = Inf,
  VAF = 0.05,
  VAFratio = 0,
  SBmethod = "SOR",
  SBscore = 3,
  maxIndelLen = 50,
  minInterval = 10,
  tagFILTER = "PASS",
  progressbar = TRUE,
  verbose = TRUE
)
```

Arguments

| | |
|-----------|--|
| maf | An MAF data frame, generated by vcfToMAF function. |
| PONfile | Panel-of-Normals files, which can be either obtained through GATK (https://gatk.broadinstitute.org/hc/en-us/articles/360035890631-Panel-of-Normals-PON-) or generated by users. Should have at least four columns: CHROM, POS, REF, ALT |
| PONformat | The format of PON file, either "vcf" or "txt". Default: "vcf" |
| panel | The sequencing panel applied on the dataset. Parameters for mutFilterQual function are set differently for different panels. Default: "Customized". Options: "MSKCC", "WES". |
| tumorDP | Threshold of tumor total depth. Default: 20 |
| normalDP | Threshold of normal total depth. Default: 10 |
| tumorAD | Threshold of tumor alternative allele depth. Default: 5 |
| normalAD | Threshold of normal alternative allele depth. Default: Inf |
| VAF | Threshold of VAF value. Default: 0.05 |
| VAFratio | Threshold of VAF ratio (tVAF/nVAF). Default: 0 |
| SBmethod | Method will be used to detect strand bias, including 'SOR' and 'Fisher'. Default: 'SOR'. SOR: StrandOddsRatio (https://gatk.broadinstitute.org/hc/en-us/articles/360041849111-StrandOddsRatio) |

| | |
|-------------|--|
| SBscore | Cutoff strand bias score used to filter variants. Default: 3 |
| maxIndelLen | Maximum length of indel accepted to be included. Default: 50 |
| minInterval | Minimum length of interval between an SNV and an indel accepted to be included. Default: 10 |
| tagFILTER | Variants with specific tag in FILTER column will be kept, set to NULL if you want to skip this filter. Default: 'PASS' |
| progressbar | Whether to show progress bar when running this function Default: TRUE |
| verbose | Whether to generate message/notification during the filtration process. Default: TRUE. |

Value

An MAF data frame after filtration for technical issue

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
maff <- mutFilterTech(maf, PONfile=system.file("extdata",
"PON_test.txt", package="CaMutQC"), PONformat="txt")
```

| | |
|---------------|----------------------|
| mutFilterType | <i>mutFilterType</i> |
|---------------|----------------------|

Description

Filter variants based on variant types

Usage

```
mutFilterType(maf, keepType = "exonic")
```

Arguments

| | |
|----------|--|
| maf | An MAF data frame, generated by vcfToMAF function. |
| keepType | A group of variant classifications will be kept, including 'exonic', 'nonsynonymous' and 'all'. Default: 'exonic'. |

Value

An MAF data frame where some variants has T tag in CaTag column for variant type filtration

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
maff <- mutFilterType(maf)
```

| | |
|--------------|---------------------|
| mutSelection | <i>mutSelection</i> |
|--------------|---------------------|

Description

Select candidate variants for cancer research.

Usage

```
mutSelection(
  maf,
  dbVAF = 0.01,
  ExAC = TRUE,
  Genomesprojects1000 = TRUE,
  ESP6500 = TRUE,
  gnomAD = TRUE,
  dbSNP = FALSE,
  keepCOSMIC = TRUE,
  keepType = "exonic",
  bedFile = NULL,
  bedHeader = FALSE,
  bedFilter = TRUE,
  progressbar = TRUE,
  verbose = TRUE
)
```

Arguments

| | |
|---------------------|---|
| maf | An MAF data frame, generated by vcfToMAF function. |
| dbVAF | Threshold of VAF of certain population for variants in database. Default: 0.01 |
| ExAC | Whether to filter variants listed in ExAC with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| Genomesprojects1000 | Whether to filter variants listed in Genomesprojects1000 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| ESP6500 | Whether to filter variants listed in ESP6500 with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| gnomAD | Whether to filter variants listed in gnomAD with VAF higher than cutoff(set in VAF parameter). Default: TRUE. |
| dbSNP | Whether to filter variants listed in dbSNP. Default: FALSE. |
| keepCOSMIC | Whether to keep variants in COSMIC even they have are present in germline database. Default: TRUE. |
| keepType | A group of variant classifications will be kept, including 'exonic', 'nonsynonymous' and 'all'. Default: 'exonic'. |
| bedFile | A file in bed format that contains region information. Default: NULL |
| bedHeader | Whether the input bed file has a header or not. Default: FALSE. |
| bedFilter | Whether to filter the information in bed file or not, which only leaves segments in Chr1-Ch22, ChrX and ChrY. Default: TRUE |

progressbar Whether to show progress bar when running this function Default: TRUE
 verbose Whether to generate message/notification during the filtration process. Default: TRUE.

Value

An MAF data frame with variants after selection.

Examples

```
maf <- vcfToMAF(system.file("extdata",
"WES_EA_T_1_mutect2.vcf", package="CaMutQC"))
mafF <- mutSelection(maf)
```

processMut

processMut

Description

Takes union or intersection on multiple MAF data frame, and return 7 important columns.

Usage

```
processMut(mafList, processMethod = "union")
```

Arguments

mafList A list of MAF data frames after going through at least one CaMutQC filtration function, and the length of the list <= 3.
 processMethod Methods for processing mutations, including "union" and "intersection". Default: "union".

Value

A data frame includes mutations after taking union or intersection.

Examples

```
maf_MuSE <- vcfToMAF(system.file("extdata/Multi-caller",
"WES_EA_T_1.MuSE.vcf", package="CaMutQC"))
maf_MuSE_f <- mutFilterCom(maf_MuSE, report=FALSE, TMB=FALSE,
PONfile=system.file("extdata", "PON_test.txt", package="CaMutQC"),
PONformat="txt")
maf_VarScan2 <- vcfToMAF(system.file("extdata/Multi-caller",
"WES_EA_T_1_varscan_filter_snp.vcf", package="CaMutQC"))
maf_VarScan2_f <- mutFilterCom(maf_VarScan2, report=FALSE, TMB=FALSE,
PONfile=system.file("extdata", "PON_test.txt", package="CaMutQC"),
PONformat="txt")
mafs <- list(maf_MuSE_f, maf_VarScan2_f)
maf_union <- processMut(mafs, processMethod="union")
```

| | |
|------------|-------------------|
| tomaftools | <i>tomaftools</i> |
|------------|-------------------|

Description

Transform a CaMutQC maf object to a maftools maf object.

Usage

```
tomaftools(
  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 | An MAF data frame, generated by <code>vcfToMAF</code> function. |
| clinicalData | Clinical data associated with each # sample/Tumor_Sample_Barcode in MAF. Could be a text file or a data.frame. Default NULL. Inherited from maftools. |
| 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. Inherited from maftools. |
| removeDuplicatedVariants | removes repeated variants in a particular sample, mapped to multiple transcripts of same Gene. See Description. Default TRUE. Inherited from maftools. |
| useAll | logical. Whether to use all variants irrespective of values in Mutation_Status. Defaults to TRUE. If FALSE, only uses with values Somatic. Inherited from maftools. |
| gisticAllLesionsFile | All Lesions file generated by gistic. e.g; all_lesions.conf_XX.txt, where XX is the confidence level. Default NULL. Inherited from maftools. |
| gisticAmpGenesFile | Amplification Genes file generated by gistic. e.g; amp_genes.conf_XX.txt, where XX is the confidence level. Default NULL. Inherited from maftools. |
| gisticDelGenesFile | Deletion Genes file generated by gistic. e.g; del_genes.conf_XX.txt, where XX is the confidence level. Default NULL. Inherited from maftools. |
| gisticScoresFile | scores.gistic file generated by gistic. Default NULL Inherited from maftools. |

| | |
|-----------|--|
| 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. Inherited from maftools. |
| cnTable | 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. Inherited from maftools. |
| isTCGA | Is input MAF file from TCGA source. If TRUE uses only first 12 characters from Tumor_Sample_Barcode. Inherited from maftools. |
| vc_nonSyn | 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. Inherited from maftools. |
| verbose | TRUE logical. Default to be talkative and prints summary. Inherited from maftools. |

Value

An maf object that can be recognized by maftools.

Examples

```
maf_CaMutQC <- vcfToMAF(system.file("extdata/Multi-caller/",
package="CaMutQC"), multiVCF=TRUE)
maf_maftools <- tomaftools(maf_CaMutQC)
```

toMesKit

toMeskit

Description

Transform a CaMutQC maf object to a MesKit maf object.

Usage

```
toMesKit(
  maf,
  clinicalFile,
  ccffile = NULL,
  nonSyn.vc = NULL,
  use.indel.ccf = FALSE,
  ccf.conf.level = 0.95
)
```

Arguments

| | |
|--------------|---|
| maf | An MAF data frame, generated by vcfToMAF function. |
| clinicalFile | A clinical data file includes Tumor_Sample_Barcode, Tumor_ID, Patient_ID. Tumor_Sample_Label is optional. |
| ccffile | A CCF file of somatic mutations. Default NULL. |
| nonSyn.vc | List of Variant classifications which are considered as non-silent. Default NULL. |

`use.indel.ccf` Whether include indels in `ccfFile`. Default FALSE.

`ccf.conf.level` The confidence level of CCF to identify clonal or subclonal. Only works when "CCF_std" or "CCF_CI_high" is provided in `ccfFile`. Default 0.95.

Value

An maf object that can be recognized by MesKit.

Examples

```
maf_CaMutQC <- vcfToMAF(system.file("extdata/Multi-caller/",
package="CaMutQC"), multiVCF=TRUE)
clin_file <- system.file("extdata", "clin.txt", package="CaMutQC")
maf_MesKit <- toMesKit(maf_CaMutQC, clinicalFile=clin_file)
```

vcfToMAF

vcfToMAF

Description

Format transformation from VCF to MAF.

Usage

```
vcfToMAF(
  vcfFile,
  multiVCF = FALSE,
  inputStrelka = FALSE,
  writeFile = FALSE,
  MAFfile = "MAF.maf",
  MAFdir = "./",
  tumorSampleName = "Extracted",
  normalSampleName = "Extracted",
  ncbiBuild = "Extracted",
  MAFcenter = ".",
  MAFstrand = "+",
  filterGene = FALSE,
  simplified = FALSE
)
```

Arguments

| | |
|---------------------------|---|
| <code>vcfFile</code> | Directory of a VCF file, or the path to several VCF files that is going to be transformed. Files should be in <code>.vcf</code> or <code>.vcf.gz</code> format. |
| <code>multiVCF</code> | Logical, whether the input is a path that leads to several VCFs that come from multi-region/sample/caller sequencing. Default: FALSE |
| <code>inputStrelka</code> | The type of variants ('INDEL' or 'SNV') in VCF file if it is from Strelka. Default: FALSE |
| <code>writeFile</code> | Whether to directly write MAF file to the disk. If FALSE, a MAF data frame will be returned. If TRUE, a MAF file will be saved. Default: FALSE. |

| | |
|------------------|--|
| MAFfile | File name of the exported MAF file, if writeFile is set as TRUE. |
| MAFdir | Directory of the exported MAF file, if writeFile is set as TRUE. |
| tumorSampleName | Name of the tumor sample(s) in the VCF file(s). If it is set as 'Extracted', tumorSampleName would be extracted automatically from the VCF file. Default: 'Extracted'. |
| normalSampleName | Name the normal sample in the VCF file. If it is set as 'Extracted', normalSampleName would be extracted automatically from the VCF file. Default: 'Extracted'. |
| ncbiBuild | The reference genome used for the alignment, which will be presented as value in 'NCBIbuild' column in MAF file. Default: 'GRCh38'. |
| MAFcenter | One or more genome sequencing center reporting the variant, which will be presented as value in 'Center' column in MAF. Default: '.'. |
| MAFstrand | Genomic strand of the reported allele, which will be presented as value in 'Strand' column in MAF file. Default: '+'. |
| filterGene | Logical. Whether to filter variants without Hugo Symbol. Default: FALSE |
| simplified | Logical. Whether to extract the first thirteen columns after converting to MAF file. Default: FALSE |

Value

A detailed MAF data frame

Examples

```
maf <- vcfToMAF(system.file("extdata", "WES_EA_T_1_mutect2.vcf",
package="CaMutQC"))
```


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