Package 'FIREVAT'

March 23, 2019

```
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Annotate VCFObj	3
CheckIfVariantRefinementIsNecessary	4
Chromosome.Names	5
ComputeZScore	5
ComputeZScoreEquiValue	6
DecimalCeiling	6
Default.Obj.Fn	7
DefaultFilterToBinary	7
EnumerateTriNucCounts	8
Euc.Exp.Weighted.Obj.Fn	8
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.1	9
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.2	9
\mathbf{J}	10
	10
	11
	11
	12
GenerateConfigObj	12
	13
	13
	14
	14
GetOptimizedSignatures	15
GetParameterLowerUpperVector	16
GetPCAWGMutSigs	16
GetPCAWGMutSigsEtiologiesColors	17
GetPCAWGMutSigsNames	17
InitializeVCF	17
	18
MutaliskParseVCFObj	18
	19
MutPatParseVCFObj	19
ParameterToBits	20
ParseConfigFile	21
	21
PCAWG.Known.Sequencing.Artifact.Signatures	22
	22
	22
	23
PerformStrandBiasAnalysis	23
· · · · · · · · · · · · · · · · · · ·	24

AnnotateVCFObj 3

	PlotMutationTypes	25
	PlotOptimizationIterations	26
	PlotSignaturesContProbs	27
	PlotTable	28
	PlotTriNucSpectrum	28
	PlotVCFStatsBoxPlots	29
	PlotVCFStatsHistograms	30
	PrepareAnnotationDB	31
	PrepareArtifactAnnotationTable	32
		32
	PrepareArtifactualMutsOptimizationIterationsPlot	33
	PrepareFilterCutoffsTable	33
	PrepareGeneticAlgorithmParametersTable	34
	PrepareIdentifiedSignaturesPlot	34
	PrepareMLEReconstructedSpectrumsPlot	35
	PrepareNucleotideSubstitutionTypesPlot	35
	PrepareObservedSpectrumsPlot	36
	PrepareOptimizationResultsTable	36
	PrepareOptimizedVCFStatisticsPlot	37
	PrepareRefinedAnnotationTable	37
	PrepareRefinedMutsOptimizationIterationsPlot	38
	PrepareRefinedStrandBiasTable	38
	PrepareResidualSpectrumsPlot	39
	PrepareTrinucleotideSpectrumsTable	39
	QueryAnnotatedVCF	40
	ReadOptimizationIterationReport	40
	ReadVCF	41
	ReportFIREVATResults	41
	RunFIREVAT	42
	RunMutalisk	44
	RunMutaliskHelper	46
		47
	TriNuc.Mutation.Type.Hex.Colors	48
	UpdateFilter	48
		49
	WriteVCF	49
		-0
Index		50
Annot	tateVCFObj AnnotateVCFObj	_

Description

Annotates a vcf.obj using df.variants.of.interest (from PrepareAnnotationDB)

Usage

```
AnnotateVCFObj(vcf.obj, df.annotation.db, columns.to.include,
  include.all.columns = FALSE)
```

Arguments

vcf.obj ReadVCF df.annotation.db

 $A \ data. frame \ from \ {\tt Prepare Annotation DB}. \ This \ data. frame \ must \ have \ the \ columns$

'CHROM', 'POS', 'REF', 'ALT'

columns.to.include

A character vector of columns to include. Note that existing columns in vcf.obj will not be affected.

include.all.columns

A boolean value. If TRUE, then annotates vcf.obj with all columns present in df.variants.of.interest. If FALSE, columns.to.include must be supplied.

Value

An annotated vcf.obj

CheckIfVariantRefinementIsNecessary

CheckIfVariantRefinementIsNecessary

Description

Checks if variant refinement is necessary by identifying mutational signatures related to sequencing artifact in the vcf.obj (set of original unrefined point mutations).

Usage

```
CheckIfVariantRefinementIsNecessary(vcf.obj, bsg, df.mut.pat.ref.sigs,
  target.mut.sigs, sequencing.artifact.mut.sigs,
  init.artifact.stop = 0.05, verbose = TRUE)
```

Arguments

vcf.obj A list from ReadVCF

bsg BSgenome.Hsapiens.UCSC object

df.mut.pat.ref.sigs

A data.frame from MutPatParseRefMutSigs

target.mut.sigs

A character vector of target mutational signatures from reference mutational signatures.

Chromosome.Names 5

sequencing.artifact.mut.sigs

A character vector of sequencing artifact mutational signatures from reference mutational signatures.

init.artifact.stop

Numeric value less than 1. If the sum of sequencing artifact weights in vcf.obj is less than or equal to this value then this function returns judgment = FALSE,

otherwise returns judgment = TRUE.

verbose

If TRUE, provides process detail. Default value is TRUE.

Value

A list with the following elements

- judgmentA boolean value
- seq.art.sigs.weights.sumA numeric value. Sum of sequencing artifact weights.

Chromosome.Names

Constant

Description

Chromosome names for FIREVAT. Chromosome names should be given in the format of "chr" + chromosome number.

Usage

Chromosome.Names

Format

An object of class character of length 25.

ComputeZScore

ComputeZScore

Description

Returns a z-score of x given a distribution of values

Usage

```
ComputeZScore(values, x)
```

Arguments

a numeric vector values a numeric value Χ

DecimalCeiling

Value

a numeric value corresponding to the z-score of x

 ${\tt Compute ZScore EquiValue}$

Compute ZS core Equi Value

Description

Returns a numeric value that is equivalent to the specified z.score in the distribution of 'values'

Usage

```
ComputeZScoreEquiValue(z.score, values)
```

Arguments

z.score numeric value values numeric vector

Value

a numeric value corresponding to the specified z.score in the 'values' distribution

DecimalCeiling

DecimalCeiling

Description

Returns the ceiling of a decimal value e.g. value = 0.15, decimal = 0.1 returns 0.2

Usage

```
DecimalCeiling(value, decimal)
```

Arguments

value numeric value (decimal)

decimal numeric value (e.g. 0.1, 0.001)

Value

a numeric value

Default.Obj.Fn 7

|--|

Description

Calculates the default objective value for FIREVAT GA optimization.

Usage

```
Default.Obj.Fn(C.refined, A.refined, C.artifactual, A.artifactual)
```

Arguments

```
C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.
```

Value

A numeric value between 0 and 1.

```
DefaultFilterToBinary Transform default filtering parameters to a binary vector
```

Description

This function transforms default filtering parameter to binary vector which can be used as a suggested solution in GA algorithm.

Usage

```
DefaultFilterToBinary(vcf.filter, params.bit.len)
```

Arguments

```
vcf.filter A list generated in MakeFilter
params.bit.len A list with bit lengths of filtering parameters which is generated from ParameterToBits
```

Value

A binary vector

Description

```
Returns C>A, C>G, C>T, T>A, T>C, T>G counts
```

Usage

EnumerateTriNucCounts(spectrum)

Arguments

spectrum a numeric vector with 96 numeric values

Details

Please note that this function assumes that 'spectrum' is sorted (i.e. $1:16 \rightarrow C>A$; $17:32 \rightarrow C>G$; $33:48 \rightarrow C>T$; $49:64 \rightarrow T>A$; $65:80 \rightarrow T>C$; $81:96 \rightarrow T>G$)

Value

a numeric vector of length 6 corresponding to the counts of each trinucleotide change (C>A, C>G, C>T, T>A, T>C, T>G)

```
Euc.Exp.Weighted.Obj.Fn
```

Euc.Exp.Weighted.Obj.Fn

Description

Calculates the Euclidean-distance of logarithmically weighted objective value for FIREVAT GA optimization.

Usage

```
Euc.Exp.Weighted.Obj.Fn(C.refined, A.refined, C.artifactual, A.artifactual)
```

Arguments

C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.

Value

A numeric value between 0 and 1.

```
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.1

Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.1
```

Description

Calculates the Euclidean-distance of logarithmically weighted objective value for FIREVAT GA optimization.

Usage

```
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.1(C.refined, A.refined, C.artifactual,
   A.artifactual)
```

Arguments

C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.

Value

A numeric value between 0 and 1.

```
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.2

Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.2
```

Description

Calculates the Euclidean-distance of logarithmically weighted objective value for FIREVAT GA optimization.

Usage

```
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.2(C.refined, A.refined, C.artifactual,
   A.artifactual)
```

Arguments

```
C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.
```

Value

A numeric value between 0 and 1.

Euc.Obj.Fn Euc.Obj.Fn

Description

Calculates the Euclidean-distance based objective value for FIREVAT GA optimization.

Usage

```
Euc.Obj.Fn(C.refined, A.refined, C.artifactual, A.artifactual)
```

Arguments

C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.

Value

A numeric value between 0 and 1.

Exp.Weighted.Obj.Fn.1 Exp.Weighted.Obj.Fn.1

Description

Calculates the exponentially weighted objective value for FIREVAT GA optimization.

Usage

```
Exp.Weighted.Obj.Fn.1(C.refined, A.refined, C.artifactual, A.artifactual)
```

Arguments

C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.

Value

A numeric value between 0 and 1.

```
Exp.Weighted.Obj.Fn.2 Exp.Weighted.Obj.Fn.2
```

Description

Calculates the exponentially weighted objective value for FIREVAT GA optimization.

Usage

```
Exp.Weighted.Obj.Fn.2(C.refined, A.refined, C.artifactual, A.artifactual)
```

Arguments

```
C.refined A numeric value between 0 and 1.

A.refined A numeric value between 0 and 1.

C.artifactual A numeric value between 0 and 1.

A.artifactual A numeric value between 0 and 1.
```

Value

A numeric value between 0 and 1.

```
\label{lem:exp.Weighted.Refined.Seq.Art.Only.Obj.Fn} Exp. Weighted. Refined. Seq. Art. Only. Obj. Fn
```

Description

Calculates the Euclidean-distance of logarithmically weighted objective value for FIREVAT GA optimization.

Usage

```
Exp.Weighted.Refined.Seq.Art.Only.Obj.Fn(C.refined, A.refined,
   C.artifactual, A.artifactual)
```

Arguments

```
C.refined A numeric value between 0 and 1.
A.refined A numeric value between 0 and 1.
C.artifactual A numeric value between 0 and 1.
A.artifactual A numeric value between 0 and 1.
```

Value

A numeric value between 0 and 1.

12 GenerateConfigObj

Description

Filter vcf based on the filter Filtering parameters are saved in config.obj Split vcf.obj into vcf.obj.filtered & vcf.obj.artifact based on vcf.filter

Usage

```
FilterVCF(vcf.obj, vcf.filter, config.obj, include.array = NULL,
  force.include = FALSE, verbose = TRUE)
```

Arguments

vcf.obj A list from ReadVCF
vcf.filter A list from MakeMuTect2Filter
config.obj A list from ParseConfigFile
include.array A boolean vector
force.include A boolean value. If TRUE, then uses 'include.array'
verbose If true, provides process detail

Value

A list with the following elements

- 1) Mutations which passed filteringvcf.obj.filtered = vcf.obj (list with data, header, genome)
- 2) Mutations which did not pass filteringvcf.obj.artifact = vcf.obj (list with data, header, genome)

GenerateConfigObj Generate config.obj by checking vcf header

Description

This function generate config.obj by checking vcf header. Users should fill in the information needed in console. In current version, only Integers & Float values can be used in config.obj for running FIREVAT.

Usage

```
GenerateConfigObj(vcf.obj, save.config = TRUE,
  config.path = "../temp/FIREVAT_configure.json")
```

GetCOSMICMutSigs 13

Arguments

vcf.obj A list from ReadVCF

save.config If true, save config.obj to config.path

config.path File path to write config.obj (json or yaml)

Value

config.obj

GetCOSMICMutSigs

GetCOSMICMutSigs

Description

Returns a data.frame of the COSMIC mutational signature reference file from the data directory

Usage

```
GetCOSMICMutSigs()
```

Value

a data.frame of the COSMIC reference mutational signatures

 ${\tt GetCOSMICMutSigsEtiologiesColors}$

GetCOSMICMutSigsNames

Description

Returns all COSMIC mutational signature etiologies and colors

Usage

```
GetCOSMICMutSigsEtiologiesColors()
```

Value

data.frame with following columns: signature, group and color.

 ${\tt GetCOSMICMutSigsNames} \quad \textit{GetCOSMICMutSigsNames}$

Description

Returns all COSMIC mutational signature names

Usage

```
GetCOSMICMutSigsNames()
```

Value

a character vector

GetGASuggestedSolutions

GetGASuggestedSolutions

Description

Computes suggested solutions

Usage

```
GetGASuggestedSolutions(vcf.obj, bsg, config.obj, lower.upper.list,
   df.mut.pat.ref.sigs, target.mut.sigs, sequencing.artifact.mut.sigs,
   objective.fn, original.muts.seq.art.weights.sum, verbose = TRUE)
```

Arguments

vcf.obj A list from ReadVCF

bsg BSgenome.Hsapiens.UCSC object

config.obj A list from ParseConfigFile

lower.upper.list

A list from GetParameterLowerUpperVector

df.mut.pat.ref.sigs

A data.frame from MutPatParseRefMutSigs

target.mut.sigs

A character vector of the target mutational signatures from reference mutational signatures.

sequencing.artifact.mut.sigs

A character vector of the sequencing artifact mutational signatures from reference mutational signatures.

```
objective.fn Objective value derivation function.
```

original.muts.seq.art.weights.sum

A numeric value. 'seq.art.sigs.weights.sum' from CheckIfVariantRefinementIs-

Necessary

verbose If TRUE, provides process detail. Default value is TRUE.

Value

A list with the following elements

- judgmentA boolean value
- seq.art.sigs.weightsA numeric value. Sum of sequencing artifact weights.

GetOptimizedSignatures

GetOptimizedSignatures

Description

This function fetches the last row from the optimization iteration log and returns the target and artifactual mutational signatures for the type of mutations ('refined' or 'artifactual')

Usage

```
GetOptimizedSignatures(data, mutations.type = "refined",
    signatures = "all")
```

Arguments

data A list of main data from RunFIREVAT

mutations.type A string for type of mutations ('refined' or 'artifact')

signatures A string ('all', 'target', 'artifact')

Value

A data.frame with the columns 'signature' and 'weight'

16 GetPCAWGMutSigs

GetParameterLowerUpperVector

GetParameterLowerUpperVector

Description

Return a lower/upper vector needed to conduct FIREVAT GA real-valued optimization.

Usage

```
GetParameterLowerUpperVector(vcf.obj, config.obj, vcf.filter,
  multiplier = 100)
```

Arguments

vcf.obj A list from ReadVCF
config.obj A list from ParseConfigFile
vcf.filter A list from MakeMuTect2Filter
multiplier A multiplier for convert fraction to integer (default = 100)

Details

vcf.obj\$data: if max(vcf.obj\$data[[param]]) < 1, then multiply multiplier to the vector

Value

A list with the elements

- lower.vector A numeric vector. Each element is the minimum value of each parameter
- upper.vector A numeric vector. Each element is the maximum value of each parameter
- · vcf.obj vcf.obj with updated data

GetPCAWGMutSigs GetPCAWGMutSigs

Description

Returns the PCAWG mutational signatures data

Usage

```
GetPCAWGMutSigs()
```

Value

a data.frame of the PCAWG mutatioanl signatures

 ${\tt GetPCAWGMutSigsEtiologiesColors}$

GetPCAWGMutSigsEtiologiesColors

Description

Returns the PCAWG mutational signatures etiologies and colors

Usage

GetPCAWGMutSigsEtiologiesColors()

Value

a data.frame with the columns 'signature', 'group', 'color'

GetPCAWGMutSigsNames GetPCAWGMutSigsNames

Description

Returns the PCAWG mutational signatures names

Usage

GetPCAWGMutSigsNames()

Value

a character vector of the PCAWG mutational signatures names

InitializeVCF

InitializeVCF

Description

Initialize VCF with FIREVAT config file This functions selects point mutations and appends filter values to vcf.obj\$data

Usage

```
InitializeVCF(vcf.obj, config.obj, verbose = TRUE)
```

Arguments

vcf.obj A list from ReadVCF
config.obj A list from ParseConfigFile
verbose If true, provides process detail

Value

A list with the following elements

- vcf.obj.filteredvcf.obj (high-quality vcf)
- vcf.obj.artifactvcf.obj (low-quality vcf)

MakeFilter

MakeFilter

Description

Creates a vcf filter from config.obj

Usage

```
MakeFilter(config.obj)
```

Arguments

config.obj

A list from ParseConfigFile (any filter with "use_in_filter" value declared as FALSE is not considered)

Value

A list with the filter parameters

MutaliskParseVCF0bj

MutaliskParseVCFObj

Description

Parses a vcf.obj and prepares it to run Mutalisk.

Usage

```
MutaliskParseVCFObj(vcf.obj)
```

Arguments

vcf.obj

A list from ReadVCF

Value

A data.frame

MutPatParseRefMutSigs MutPatParseRefMutSigs

Description

Parses a df.ref.mut.sigs and prepares it to run Mutational Patterns.

Usage

```
MutPatParseRefMutSigs(df.ref.mut.sigs, target.mut.sigs,
    signature.start.column.index = 4,
    mutation.type.header = "SomaticMutationType")
```

Arguments

```
df.ref.mut.sigs

A data.frame of reference mutational signatures

target.mut.sigs

A character vector of target mutational signatures names

signature.start.column.index

= An integer value (e.g. column index corresponding to 'SBS1')

mutation.type.header

= A string value (name of header corresponding to column containing 'A[C>A]A' data))
```

Value

A data.frame of the format deconstructSigs::signatures.cosmic

MutPatParseVCFObj MutPatParseVCFObj

Description

Parses a vcf.obj and prepares it to run Mutational Patterns.

Usage

```
MutPatParseVCFObj(vcf.obj, bsg, sample.id = "sample")
```

20 ParameterToBits

Arguments

vcf.obj A list from ReadVCF

bsg BSgenome.Hsapiens.UCSC.hg19::BSgenome.Hsapiens.UCSC.hg19 or BSgenome.Hsapiens.UCSC.hg38

sample.id A string value

Value

A data frame with the column sample id and row names corresponding to 96 substitution types

ParameterToBits ParameterToBits

Description

Calculate the number of bits needed to conduct FIREVAT GA binary optimization.

Usage

```
ParameterToBits(vcf.obj, config.obj, vcf.filter, multiplier = 100)
```

Arguments

vcf.obj A list from ReadVCF

config.obj A list from ParseConfigFile

vcf.filter A list from MakeMuTect2Filter

multiplier A multiplier for convert fraction to integer (default = 100)

Details

vcf.obj\$data: if max(vcf.obj\$data[[param]]) < 1, then multiply multiplier to the vector

Value

A list with the elements

- params.bit.lenA numeric vector. Each element is the bit length of each parameter value
- vcf.objA vcf.obj (ReadVCF) with updated data

ParseConfigFile 21

ParseConfigFile

Parse Config File

Description

This function returns config.obj from JSON or YAML config file. - Check if the config file is in JSON format or YAML format - Return config.obj

Usage

```
ParseConfigFile(config.path, verbose = TRUE)
```

Arguments

config.path A string for config file path verbose If true, provides process detail

Value

```
config.obj: list of parameters
```

Examples

```
## Not run:
ParseConfigFile("example.variant.caller.json")
ParseConfigFile("example.variant.caller.json", verbose=False)
## End(Not run)
```

```
PCAWG.All.Sequencing.Artifact.Signatures
                       Constant
```

Description

PCAWG mutational signatures reported to be associated with sequencing artifacts

Usage

```
PCAWG.All.Sequencing.Artifact.Signatures
```

Format

An object of class character of length 17.

PCAWG.Known.Sequencing.Artifact.Signatures

Constant

Description

PCAWG mutational signatures reported to be associated with sequencing artifacts

Usage

PCAWG.Known.Sequencing.Artifact.Signatures

Format

An object of class character of length 1.

PCAWG.Likely.Sequencing.Artifact.Signatures

Constant

Description

PCAWG mutational signatures reported to be associated with sequencing artifacts

Usage

PCAWG.Likely.Sequencing.Artifact.Signatures

Format

An object of class character of length 5.

 ${\it PCAWG.Possible.Sequencing.Artifact.Signatures} \\ {\it Constant}$

Description

PCAWG mutational signatures reported to be associated with sequencing artifacts

Usage

PCAWG.Possible.Sequencing.Artifact.Signatures

Format

An object of class character of length 11.

```
{\it PCAWG.Target.Mutational.Signatures} \\ {\it Constant}
```

Description

PCAWG target mutational signatures reported to be unrelated to sequencing artifacts

Usage

```
PCAWG. Target. Mutational. Signatures
```

Format

An object of class character of length 49.

PerformStrandBiasAnalysis

PerformStrandBiasAnalysis

Description

Performs strand bias analysis

Usage

```
PerformStrandBiasAnalysis(vcf.obj, ref.forward.strand.var,
  ref.reverse.strand.var, alt.forward.strand.var, alt.reverse.strand.var,
  perform.fdr.correction = TRUE, fdr.correction.method = "BH")
```

Arguments

24 PlotMutaliskResults

Value

An updated vcf.obj

Description

Plots Mutalisk results

Usage

```
PlotMutaliskResults(mutalisk.results, signatures, trinuc.max.y,
    trinuc.min.y, mut.type.max.y, title)
```

Arguments

```
mutalisk.results
A list obtained from RunMutalisk
signatures A character vector of mutational signatures names
trinuc.max.y A numeric value (maximum y-axis value)
trinuc.min.y A numeric value (minimum y-axis value)
mut.type.max.y A numeric value
title A string value
```

Value

A ggplot object

Examples

PlotMutationTypes 25

PlotMutationTypes

PlotMutationTypes

Description

Plots a horizontal barplot of mutation types

Usage

```
PlotMutationTypes(mutation.types = c("C>A", "C>G", "C>T", "T>A", "T>C",
   "T>G"), mutation.types.values, mutation.types.colors, max.y.val, title,
   convert.to.percentage = T, show.legend = T, font.size.small = 8,
   font.size.med = 14, plot.margin = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
```

Arguments

```
mutation.types \;\; Mutation \; types; \; Default = c("C>A", "C>G", "C>T", "T>A", "T>C", "T>G")
mutation.types.values
                  Mutation count for each mutation type
mutation.types.colors
                  A color vector for indicating mutation types
max.y.val
                  y axis maximum value
                  Plot title
title
convert.to.percentage
                  if True convert y values to percentage (x 100); Default = T
                  If True, show legend; Default = T
show.legend
font.size.small
                  Small font size; Default = 8
font.size.med
                  Medium font size; Default = 14
                  Margin vector for drawing plot; Default = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
plot.margin
```

Value

A ggplot object

Examples

```
plot.margin = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
print(p)
## End(Not run)
```

PlotOptimizationIterations

PlotOptimizationIterations

Description

Plots multiple scatter plots into one figure

Usage

```
PlotOptimizationIterations(df, columns.to.plot, x.axis.var, x.axis.title,
  x.max, save.file, title, y.axis.title = "", y.max = 1,
  point.size = 1, connect.dots = T, plot.legend = T,
  legend.ncol = 1, font.size.med = 14, font.size.large = 16,
  plot.margin = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
```

Arguments

```
df
                  A data.frame (from reading "FIREVAT_Optimization_Logs.tsv")
columns.to.plot
                  A character vector (of column names to plot)
x.axis.var
                  x axis variable
x.axis.title
                  x axis title
                  x axis maximum value
x.max
save.file
                  Filename (including full path) to which the plot will be saved
title
                  Plot title
                  y axis title; Default = ""
y.axis.title
                  y axis maximum value; Default = 1
y.max
                  Point size: Default = 1
point.size
connect.dots
                  If True draws dots for each iteration; Default = True
plot.legend
                  If True write legend of plot; Default = T
legend.ncol
                  legend.n Default = 1
font.size.med
                  Medium font size; Default = 14
font.size.large
                  Large font size; Default = 16
                  Margin vector for plot; Default = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
plot.margin
```

Value

A ggplot object

```
PlotSignaturesContProbs
```

PlotSignaturesContProbs

Description

Plots a horizontal barplot of identified mutational signatures

Usage

```
PlotSignaturesContProbs(df.identified.mut.sigs, df.ref.sigs.groups.colors,
  title, convert.to.percentage = T, font.size.small = 8,
  font.size.med = 14, plot.margin = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
```

Arguments

```
df.identified.mut.sigs
A data.frame of identified mutational signatures

df.ref.sigs.groups.colors
A data.frame with 'signature', 'group', and 'color' columns

title
Plot title
convert.to.percentage
If true, convert y values to percentage (x 100); Default = T,

font.size.small
Small font size; Default = 8,

font.size.med
Medium font size; Default = 14,

plot.margin
Margin vector for drawing plot; Default = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))
```

Value

A ggplot object

Examples

```
## Not run:
    g <- PlotSignaturesContProbs(sigs = c(mutalisk.results$identified.mut.sigs),
    sigs.probs = c(mutalisk.results$identified.mut.sigs.probs),
    df.ref.sigs.groups.colors = GetPCAWGMutSigsEtiologiesColors())
    print(g)
## End(Not run)</pre>
```

PlotTriNucSpectrum

PlotTable

PlotTable

Description

Plots basic statistics table

Usage

```
PlotTable(df, padding = 20, font.size = 14)
```

Arguments

df = A data.frame where the first column is header and the second column is data

value

padding size; Default = 20

font.size Font size; Default = 14

Value

A plot

PlotTriNucSpectrum

PlotTriNucSpectrum

Description

Plots the spectrum of 96 trinucleotide distribution (C>A, C>G, C>T, T>A, T>C, T>G) Please note that this function assumes that both sub.types and spectrum are sorted in the following order: C>A, C>G, C>T, T>A, T>C, T>G

Usage

```
PlotTriNucSpectrum(sub.types, spectrum, max.y.val, min.y.val, y.axis.title,
  draw.top.strip = T, draw.x.axis.labels = T, draw.y.axis.labels = T,
  draw.y.axis.title = T, font.size.small = 8, font.size.med = 14,
  plot.margin.top = 0.5, plot.margin.bottom = 0.5,
  plot.margin.left = 0.5, plot.margin.right = 0.5, title)
```

PlotVCFStatsBoxPlots 29

Arguments

```
A character vector (types of 96 trinucleotide substitutions)
sub.types
                  A numeric vector (96 elements)
spectrum
                 y axis maximum value
max.y.val
min.y.val
                 y axis minimum value
y.axis.title
                  y axis title
draw.top.strip If True then draws top strip; Default = T
draw.x.axis.labels
                  If True then draws x axis labels; Default = T
draw.y.axis.labels
                  If True then draws y axis labels; Default = T
draw.y.axis.title
                  If True then draws y axis title; Default = T
font.size.small
                  Small font size; Default = 8
font.size.med
                 Medium font size; Default = 14
plot.margin.top
                  Top margin; Default = 0.5
plot.margin.bottom
                  Bottom margin; Default = 0.5
plot.margin.left
                 Left margin; Default = 0.5
plot.margin.right
                 Right margin; Default = 0.5
title
                  Plot title
```

Value

A ggplot object

Description

Plots multiple (original, refined, artifact vcf) boxplots for single filter parameter

Usage

```
PlotVCFStatsBoxPlots(original.vcf.stat.values, refined.vcf.stat.values,
  artifact.vcf.stat.values, xlab, axis.font.size = 10,
  label.font.size = 10, title.font.size = 12)
```

Arguments

```
A numeric vector corresponding to the original vcf.obj values of single filter parameter

refined.vcf.stat.values

A numeric vector corresponding to the refined vcf.obj values of single filter parameter

artifact.vcf.stat.values

A numeric vector corresponding to the artifact vcf.obj values of single filter parameter

xlab

A string value (x-axis label)

axis.font.size

An integer value (axis font size)

label.font.size

An integer value (label font size)
```

Value

A ggboxplot

PlotVCFStatsHistograms

PlotVCFStatsHistograms

An integer value (title font size)

Description

Plots multiple VCF stats histograms into one figure

Usage

```
PlotVCFStatsHistograms(plot.values, x.axis.labels, stat.y.max.vals,
    stat.x.max.vals, sample.id, save.file, title, cutoff.values,
    plot.boxplot = F, plot.cutoff.line.color = "#D4012E",
    plot.cutoff.value.lines = F, bin.width = 1, ncol = 4, nrow = 3,
    font.size.med = 10, font.size.large = 12, plot.margin = unit(c(0.5,
    0.5, 0.5, 0.5), "cm"))
```

Arguments

```
plot.values A list of multiple numeric vectors x.axis.labels A character vector of x axis labels stat.y.max.vals
```

A numeric vector of max y-axis values

PrepareAnnotationDB 31

stat.x.max.vals

A numeric vector of max x-axis values

sample.id A string value of sample ID

save.file A string value of file to which the resulting plot will be saved

title A string value of plot title

cutoff.values A numeric vector of cutoff values plot.boxplot A boolean value (default = False)

plot.cutoff.line.color

A hex string value (default = "#D4012E")

plot.cutoff.value.lines

A boolean value (default = False)

bin.width An integer value (default = 1; histogram bin width)

ncol An integer value (default = 4; ggarrange ncol)

nrow An integer value (default = 3; ggarrange nrow)

font.size.med An integer value (default = 10)

font.size.large

An integer value (default = 12)

plot.margin A list (default = unit(c(0.5, 0.5, 0.5, 0.5), "cm"))

Value

A list with the following elements

- f = A ggarrange object
- graphs = A list of length 3; each element is a ggplot histogram

PrepareAnnotationDB

Description

Prepares df.genes.of.interest from a vcf.obj (ReadVCF) of COSMIC or ClinVar vcf for AnnotateVCF0bj

Usage

PrepareAnnotationDB(annotation.vcf.obj)

Arguments

```
annotation.vcf.obj
```

vcf.obj of COSMIC or ClinVar vcf file

Value

A data.frame of annotation.vcf.obj

PrepareArtifactAnnotationTable

Prepare Artifact Annotation Table

Description

Prepares artifactual mutations annotation (filtered, queried) table

Usage

PrepareArtifactAnnotationTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

 ${\tt Prepare Artifact Strand Bias Table}$

Prepare Artifact Strand Bias Table

Description

Prepares artifactual mutations strand biased variants table

Usage

PrepareArtifactStrandBiasTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

 $\label{lem:prepareArtifactualMutsOptimizationIterationsPlot} PrepareArtifactual \texttt{MutsOptimizationIterationsPlot}$

 $\label{lem:prepareArtifactual MutsOptimization Iterations Plot} Prepare Artifactual MutsOptimization Iterations Plot$

Description

Prepares artifactual mutations optimization iterations plot

Usage

 $\label{lem:prepareArtifactualMutsOptimizationIterationsPlot(data)} PrepareArtifactualMutsOptimizationIterationsPlot(data)$

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggplot object

 ${\tt PrepareFilterCutoffsTable}$

Prepare Filter Cutoffs Table

Description

Prepares filter cutoffs table for reporting

Usage

PrepareFilterCutoffsTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

 ${\tt Prepare Genetic Algorithm Parameters Table}$

Prepare Genetic Algorithm Parameters Table

Description

Prepares Genetic Algorithm parameters table

Usage

 ${\tt Prepare Genetic Algorithm Parameters Table (data)}$

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

 ${\tt PrepareIdentifiedSignaturesPlot}$

Prepare Identified Signatures Plot

Description

Prepares identified signatures plot for reporting

Usage

PrepareIdentifiedSignaturesPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggarrange object

 ${\tt Prepare MLERe constructed Spectrum sPlot}$

Prepare MLE Reconstructed Spectrums Plot

Description

Prepares MLE reconstructed spectrums plot

Usage

PrepareMLEReconstructedSpectrumsPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggarrange object

 $\label{lem:prepareNucleotideSubstitutionTypesPlot} PrepareNucleotideSubstitutionTypesPlot$

Description

Prepares nucleotide substitution types plot

Usage

PrepareNucleotideSubstitutionTypesPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggarrange object

PrepareObservedSpectrumsPlot

 ${\it Prepare Observed Spectrums Plot}$

Description

Prepares observed spectrums plot

Usage

PrepareObservedSpectrumsPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggarrange object

 ${\tt PrepareOptimizationResultsTable}$

Prepare Optimization Results Table

Description

Prepares optimization results table

Usage

PrepareOptimizationResultsTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

PrepareOptimizedVCFStatisticsPlot

Prepare Optimized VCF Statistics Plot

Description

Prepares optimized VCF statistics plot

Usage

PrepareOptimizedVCFStatisticsPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggarrange object

 ${\tt Prepare Refined Annotation Table}$

Prepare Refined Annotation Table

Description

Prepares refined mutations annotation (filtered, queried) table

Usage

PrepareRefinedAnnotationTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

 $\label{lem:prepareRefinedMutsOptimizationIterationsPlot} Prepare Refined MutsOptimization Iterations Plot$

Description

Prepares refined mutations optimization iterations plot

Usage

PrepareRefinedMutsOptimizationIterationsPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggplot object

 ${\tt Prepare Refined Strand Bias Table}$

Prepare Refined Strand Bias Table

Description

Prepares refined mutations strand biased variants table

Usage

PrepareRefinedStrandBiasTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

 ${\tt PrepareResidualSpectrumsPlot}$

Prepare Residual Spectrums Plot

Description

Prepares residual spectrums plot

Usage

PrepareResidualSpectrumsPlot(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A ggarrange object

 ${\tt PrepareTrinucleotideSpectrumsTable}$

Prepare Trinucle ot ide Spectrums Table

Description

Prepares trinucleotide spectrums table

Usage

PrepareTrinucleotideSpectrumsTable(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame

QueryAnnotatedVCF

FilterAnnotatedVCF

Description

Annotates a vcf.obj using df.variants.of.interest (from (PrepareAnnotationDB)

Usage

```
QueryAnnotatedVCF(vcf.obj.annotated, filter.key.value.pairs,
  filter.condition = "AND")
```

Arguments

```
vcf.obj.annotated
```

AnnotateVCF0bj

filter.key.value.pairs

A list with the key as the column name and value as the filtering values. E.g. list("CLNSIG" = c("Pathogenic", "Pathogenic/Likely_pathogenic"))

filter.condition

'AND' or 'OR'.

Value

A vcf.obj

 ${\tt ReadOptimizationIterationReport}$

ReadOptimizationIterationReport

Description

Read optimization iteration report

Usage

ReadOptimizationIterationReport(data)

Arguments

data

A list of elements returned from RunFIREVAT

Value

A data.frame of FIREVAT optimization logs

ReadVCF 41

ReadVCF

ReadVCF

Description

Reads a .vcf file

Usage

```
ReadVCF(vcf.file, genome = "hg19", split.info = FALSE,
   check.chromosome.name = TRUE)
```

Arguments

vcf.file (full path of a .vcf file) genome ('hg19' or 'hg38')

split.info A boolean value. If TRUE, then makes the INFO column in the vcf as a separate

column. Default value is FALSE.

check.chromosome.name

A boolean value. If TRUE, then check whether converts 'MT' to 'M' and adds

'chr' to the CHROM column. Default value is TRUE.

Value

A list with elements 'data', 'header', 'genome'

```
ReportFIREVATResults ReportFIREVATResults
```

Description

Reports FIREVAT results in html format (generated from Rmd)

Usage

```
ReportFIREVATResults(data)
```

Arguments

data

A list of main data from RunFIREVAT

Value

An updated data list

42 RunFIREVAT

RunFIREVAT

RunFIREVAT

Description

Runs FIREVAT using configuration data. Filters point mutations in the user-specified vcf file based on mutational signature identification and outputs the refined and artifact vcf files as well as metadata related to the refinement process.

Usage

```
RunFIREVAT(vcf.file, vcf.file.genome, config.file, df.ref.mut.sigs,
 target.mut.sigs, sequencing.artifact.mut.sigs, num.cores, output.dir,
 mode = "ga", init.artifact.stop = 0.05,
 objective.fn = Default.Obj.Fn, use.suggested.soln = TRUE,
 ga.type = "real-valued", ga.pop.size = 200, ga.max.iter = 200,
 ga.run = 50, ga.pmutation = 0.25, mutalisk = TRUE,
 mutalisk.method = "all", mutalisk.random.sampling.count = 20,
 mutalisk.random.sampling.max.iter = 10,
 perform.strand.bias.analysis = TRUE,
 strand.bias.perform.fdr.correction = TRUE,
 strand.bias.fdr.correction.method = "BH",
 ref.forward.strand.var = NULL, ref.reverse.strand.var = NULL,
 alt.forward.strand.var = NULL, alt.reverse.strand.var = NULL,
 annotate = TRUE, df.annotation.db = NULL,
 annotated.columns.to.display = NULL,
 annotation.filter.key.value.pairs = NULL,
 annotation.filter.condition = "AND", write.vcf = TRUE,
 report = TRUE, save.rdata = TRUE, save.tsv = TRUE,
 report.format = "html", verbose = TRUE)
```

Arguments

vcf. file String value corresponding to input .vcf file. Please provide the full path.

vcf.file.genome

Genome assembly of the input .vcf file. The value should be eitehr 'hg19' or 'hg38'.

config.file

String value corresponding to input configuration file. For more details please refer to ...

df.ref.mut.sigs

A data.frame of the reference mutational signatures

target.mut.sigs

A character vector of the target mutational signatures from reference mutational signatures.

sequencing.artifact.mut.sigs

A character vector of the sequencing artifact mutational signatures from reference mutational signatures.

RunFIREVAT 43

num. cores Number of cores to allocate

output.dir String value of the desired output directory

mode String value. The value should be either 'ga' or 'manual'.

init.artifact.stop

Numeric value less than 1. If the sum of sequencing artifact weights in the user-specified original VCF file (i.e. vcf.file) is less than or equal to this value then FIREVAT does not perform variant refinement. Default value is 0.05. Note that this option does not apply if 'mode' is 'manual'.

objective.fn Objective value derivation function. Default: Default.Obj.Fn.

use.suggested.soln

Boolean value. If TRUE, then FIREVAT passes the default values of filter variables declared as 'use_in_filter' in the config file to the 'suggestions' parameter of the Genetic Algorithm package. If FALSE, then FIREVAT supplies NULL to the GA package 'suggestions' parameter. FIREVAT also computes baseline performance of each filter variable and uses fittest population from each variable as a suggested solution.

ga. type String value. The value should be either 'binray' or 'real-valued'.

ga.pop.size Integer value of the Genetic Algorithm 'population size' parameter. Default: 200. This value should be set based on the number of filter parameters. Recom-

mendation: 40 per filter parameter.

ga.max.iter Integer value of the Genetic Algorithm 'maximum iterations' parameter. Dde-

fault: 200. This value should be set based on the number of filter parameters.

Recommendation: same as 'ga.pop.size'.

ga.run Integer value of the Genetic Algorithm 'run' parameter. Default: 50. This value

should be set based on the 'ga.max.iter' parameter. Recommendation: 25 per-

cent of 'ga.max.iter'.

ga.pmutation Float value of the Genetic Algorithm 'mutation probability' parameter. Default:

0.25.

mutalisk If TRUE, confirm mutational signature analysis with Mutalisk. Default: TRUE.

mutalisk.method

Mutalisk signature identification method. Default: 'random.sampling'. The value can be either 'all' or 'random.sampling'. 'all' uses all target.mut.sigs to identify mutational signatures. 'random.sampling' randomly samples from target.mut.sigs to identify mutational signatures.

mutalisk.random.sampling.count

Mutalisk random sampling count. Default: 20. The number of signatures to sample from target.mut.sigs

mutalisk.random.sampling.max.iter

Mutalisk random sampling maximum iteration. Default: 10. The number of times Mutalisk randomly samples from target.mut.sigs before determining the candidate signatures.

perform.strand.bias.analysis

If TRUE, then performs strand bias analysis.

strand.bias.perform.fdr.correction

If TRUE, then performs false discovery rate correction for strand bias analysis.

44 RunMutalisk

strand.bias.fdr.correction.method

A string value. Default value is 'BH'. Refer to 'p.adjust()' function method.

ref.forward.strand.var

A string value.

ref.reverse.strand.var

A string value,

alt.forward.strand.var

A string value,

alt.reverse.strand.var

A string value,

annotate A boolean value. Default value is TRUE.

df.annotation.db

A data.frame. Please refer to PrepareAnnotationDB

annotated.columns.to.display

A character vector.

annotation.filter.key.value.pairs

A list.

annotation.filter.condition

'AND' or 'OR'.

write.vcf If TRUE, write original/refined/artifact vcfs. Default: TRUE.

report If TRUE, generate report. Default: TRUE.
save.rdata If TRUE, save rdata. Default: TRUE.
save.tsv If TRUE, save tsv. Default: TRUE.

report. format The format of FIREVAT report. We currently only support 'html'.

verbose If TRUE, provides process detail. Default: TRUE.

Value

A list with the following elements

- f = A ggarrange object
- graphs = A list of length 3; each element is a ggplot histogram

RunMutalisk

RunMutalisk

Description

Identifies mutational signatures using Mutalisk

Usage

```
RunMutalisk(vcf.obj, df.ref.mut.sigs, target.mut.sigs,
  random.sampling.candidate.mut.sigs = c(), method = "random.sampling",
  n.sample = 20, n.iter = 10, verbose = TRUE)
```

RunMutalisk 45

Arguments

A list (from firevat vcf::ReadVCF) vcf.obj

df.ref.mut.sigs

A data.frame of reference mutational signatures

target.mut.sigs

A character vector of target mutational signatures names to identify from

random.sampling.candidate.mut.sigs

A character vector of mutational signatures names that gets appended to the list

of candidate mutational signatures so that these are always considered.

method A string value (must be either 'random.sampling' or 'all'). The method 'ran-

> dom.sampling' samples (without replacement) 'n.sample' number of signatures 'n.iter' number of times and runs the candidate signatures one last time. The

method 'all' uses all target.mut.sigs

An integer value ('random.sampling' method parameter) Number of signatures n.sample

to choose for each iteration of random sampling).

n.iter An integer value ('random.sampling' method parameter). Number of iterations

to perform random sampling.

If true, provides process details verbose

Value

A list with the following elements

- num.point.mutationsAn integer value count of total point mutations
- sub.typesA character vector of length 96
- sub.types.spectrumA numeric vector of length 96
- num.mut.sigsAn integer value (count of unique mutational signatures identified)
- · identified.mut.sigsA character vector where each element is a mutational signature identified
- · identified.mut.sigs.probsA numeric vector where each element is the weight of mutational signature identified. The ordering follows identified.mut.sigs
- identified.mut.sigs.spectrumA numeric vector of length 96
- · residualsA numeric vector of length 96
- rssA numeric value (residual sum of squares)
- · cos.sim.scoreA numeric value (cosine similarity score between observed mutational spectrum and reconstructed mutational signatures)
- all.models.sigsA list where each element is a model; a model is a list of signatures identified)
- all.models.sigs.probsA list where each element is a model; a model is a list of contribution probabilities
- all.models.cos.sim.scoresA list where each element is a model; a model is a list of cosine similarity socres

46 RunMutaliskHelper

RunMutaliskHelper

RunMutaliskHelper

Description

Helper function for RunMutalisk

Usage

```
RunMutaliskHelper(vcf.trinucleotide.data, df.ref.mut.sigs, target.mut.sigs)
```

Arguments

```
vcf.trinucleotide.data
A data.frame (from firevat_mutalisk::MutaliskParseVCFObj)

df.ref.mut.sigs
A data.frame of reference mutational signatures

target.mut.sigs
A character vector of target mutational signatures names
```

Value

A list with the following elements

- num.point.mutationsAn integer value count of total point mutations
- sub.typesA character vector of length 96
- sub.types.spectrumA numeric vector of length 96
- num.mut.sigsAn integer value (count of unique mutational signatures identified)
- · identified.mut.sigsA character vector where each element is a mutational signature identified
- identified.mut.sigs.probsA numeric vector where each element is the weight of mutational signature identified. The ordering follows identified.mut.sigs
- identified.mut.sigs.spectrumA numeric vector of length 96
- residualsA numeric vector of length 96
- rssA numeric value (residual sum of squares)
- cos.sim.scoreA numeric value (cosine similarity score between observed mutational spectrum and reconstructed mutational signatures)
- all.models.sigsA list where each element is a model; a model is a list of signatures identified)
- all.models.sigs.probsA list where each element is a model; a model is a list of contribution probabilities
- all.models.cos.sim.scoresA list where each element is a model; a model is a list of cosine similarity socres

RunMutPat 47

RunMutPat RunMutPat

Description

Identifies mutational signatures using Mutational Patterns

Usage

```
RunMutPat(mut.pat.input, df.mut.pat.ref.sigs, target.mut.sigs,
  verbose = TRUE)
```

Arguments

Value

A list with the following elements

- tumor.mutation.types.spectrumA numeric vector of length 96 'observed' spectrum
- identified.mutation.types.spectrumA numeric vector of length 96 'identified' spectrum
- residuals A numeric vector of length 96 residuals
- mutation.typesA character vector of length 96
- · identified.mut.sigsA character vector where each element is a mutational signature identified
- identified.mut.sigs.contribution.weightsA numeric vector where each element is the weight of mutational signature identified. The ordering follows identified.mut.sigs
- cosine.similarity.scoreA numeric value

Examples

```
## Not run:
vcf.obj <- ReadVCF(vcf.file = "../data/sample/HNT-082-BT.final.call.vcf", genome = "hg19")
df.ref.mut.sigs <- GetPCAWGMutSigs()
target.mut.sigs <- GetPCAWGMutSigsNames()
RunMutPat(vcf.obj = vcf.obj,
df.ref.mut.sigs = df.ref.mut.sigs,
target.mut.sigs = target.mut.sigs)
## End(Not run)</pre>
```

48 UpdateFilter

```
\label{total:colors} TriNuc. \texttt{Mutation.Type.Hex.Colors} \\ \textit{Constant}
```

Description

Hex codes for the mutation types (for plotting purposes)

Usage

```
TriNuc.Mutation.Type.Hex.Colors
```

Format

An object of class character of length 6.

UpdateFilter

UpdateFilter

Description

Update filter based on optim parameter values

Usage

```
UpdateFilter(vcf.filter, param.values)
```

Arguments

vcf.filter A list from MakeFilterFromConfig

param. values A numeric vector contains filtering value (same length with length(vcf.config.filter))

Value

Updated vcf.filter (list)

WriteFIREVATResultsToTSV

WriteFIREVATResultsToTSV

Description

Writes FIREVAT results to a csv file

Usage

```
WriteFIREVATResultsToTSV(firevat.results)
```

Arguments

firevat.results

List returned from RunFIREVAT

WriteVCF

WriteVCF

Description

Writes a vcf.obj to a .vcf file

Usage

```
WriteVCF(vcf.obj, save.file)
```

Arguments

vcf.obj (from the function ReadVCF) save.file (full path including filename)

Index

```
*Topic datasets
                                               GetCOSMICMutSigs, 13
    Chromosome. Names, 5
                                               GetCOSMICMutSigsEtiologiesColors, 13
    PCAWG.All.Sequencing.Artifact.Signatures,GetCOSMICMutSigsNames, 14
                                               GetGASuggestedSolutions, 14
    PCAWG.Known.Sequencing.Artifact.SignatureSetOptimizedSignatures, 15
                                               GetParameterLowerUpperVector, 16
    PCAWG.Likely.Sequencing.Artifact.Signatur@stPCAWGMutSigs, 16
                                               GetPCAWGMutSigsEtiologiesColors, 17
    PCAWG.Possible.Sequencing.Artifact.SignatQetBCAWGMutSigsNames, 17
                                               InitializeVCF, 17
    PCAWG. Target. Mutational. Signatures,
                                               MakeFilter, 7, 18
    TriNuc.Mutation.Type.Hex.Colors,
                                               MutaliskParseVCFObj, 18
                                               MutPatParseRefMutSigs, 19, 47
AnnotateVCFObj, 3, 31, 40
                                               MutPatParseVCFObj, 19, 47
CheckIfVariantRefinementIsNecessary, 4
                                               ParameterToBits, 7, 20
Chromosome. Names, 5
                                               ParseConfigFile, 21
ComputeZScore, 5
                                               PCAWG.All.Sequencing.Artifact.Signatures,
ComputeZScoreEquiValue, 6
                                               PCAWG.Known.Sequencing.Artifact.Signatures,
DecimalCeiling, 6
Default.Obj.Fn, 7
                                               PCAWG.Likely.Sequencing.Artifact.Signatures,
DefaultFilterToBinary, 7
                                               PCAWG.Possible.Sequencing.Artifact.Signatures,
EnumerateTriNucCounts, 8
Euc.Exp.Weighted.Obj.Fn, 8
                                               PCAWG. Target. Mutational. Signatures, 23
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.1,
                                               PerformStrandBiasAnalysis, 23
                                               PlotMutaliskResults, 24
Euc.Exp.Weighted.Seq.Art.Only.Obj.Fn.2,
                                               PlotMutationTypes, 25
                                               PlotOptimizationIterations, 26
Euc. Obj. Fn, 10
                                               PlotSignaturesContProbs, 27
Exp.Weighted.Obj.Fn.1, 10
                                               PlotTable, 28
Exp.Weighted.Obj.Fn.2, 11
                                               PlotTriNucSpectrum, 28
Exp.Weighted.Refined.Seq.Art.Only.Obj.Fn,
                                               PlotVCFStatsBoxPlots, 29
        11
                                               PlotVCFStatsHistograms, 30
                                               PrepareAnnotationDB, 3, 4, 31, 40, 44
FilterVCF. 12
                                               PrepareArtifactAnnotationTable, 32
GenerateConfigObj, 12
                                               PrepareArtifactStrandBiasTable, 32
```

INDEX 51

```
PrepareArtifactualMutsOptimizationIterationsPlot,
PrepareFilterCutoffsTable, 33
PrepareGeneticAlgorithmParametersTable,
        34
{\tt PrepareIdentifiedSignaturesPlot}, 34
PrepareMLEReconstructedSpectrumsPlot,
PrepareNucleotideSubstitutionTypesPlot,
        35
PrepareObservedSpectrumsPlot, 36
PrepareOptimizationResultsTable, 36
PrepareOptimizedVCFStatisticsPlot, 37
PrepareRefinedAnnotationTable, 37
{\tt Prepare Refined Muts Optimization Iterations Plot},
{\tt Prepare Refined Strand Bias Table,\,38}
PrepareResidualSpectrumsPlot, 39
PrepareTrinucleotideSpectrumsTable, 39
QueryAnnotatedVCF, 40
ReadOptimizationIterationReport, 40
ReadVCF, 4, 13, 20, 23, 31, 41
ReportFIREVATResults, 41
RunFIREVAT, 15, 32–41, 42
RunMutalisk, 24, 44
RunMutaliskHelper, 46
RunMutPat, 47
{\tt TriNuc.Mutation.Type.Hex.Colors,48}
UpdateFilter, 48
WriteFIREVATResultsToTSV, 49
WriteVCF, 49
```