Package 'SynSigEval'

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```
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     Examine and evaluate the output of mutational signature analysis computational approaches.
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```

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 ${\tt CopyBestSignatureAnalyzerResult}$

Find the SignatureAnalyzer results directory with the best results and make a copy of it as sa.results.dir/best.run/

Description

Find the SignatureAnalyzer results directory with the best results and make a copy of it as sa.results.dir/best.run/

```
CopyBestSignatureAnalyzerResult(
  sa.results.dir,
  verbose = FALSE,
  overwrite = FALSE
)
```

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Arguments

```
sa.results.dir See BestSignatureAnalyzerResult
verbose See BestSignatureAnalyzerResult
overwrite If TRUE overwrite existing "best.run"
```

Value

The path of the best directory that was copied as a string, with the list directories examined as the attribute run. directories.

CreateEMuOutput

Prepare input file for EMu from a EMu formatted catalog file.

Description

Prepare input file for EMu from a EMu formatted catalog file.

Usage

```
CreateEMuOutput(
  catalog,
  out.dir = paste0(dirname(catalog), "/ExtrAttr/EMu.results"),
  overwrite = FALSE
)
```

Arguments

catalog a catalog in ICAMS format. It can be a .csv file, or a matrix or data.frame.

Usually, it refers to "ground.truth.syn.catalog.csv".

out.dir Directory that will be created for the output; abort if it already exists. Usu-

ally, the out.dir will be a EMu.results folder directly under the folder storing

catalog.

overwrite If TRUE, overwrite existing output

Details

Creates folder named EMu.results containing catalogs in EMu-formatted catalogs: Rows are signatures; the first column is the name of the mutation type, while the remaining columns are samples (tumors). These EMu-formatted catalogs will the input when running EMu program later on compiled binary.

Value

invisible(catalog), original catalog in EMu format

 $\label{lem:continuous} \textit{CreatehelmsmanOutput} \quad \textit{Prepare input file for helmsman from a helmsman formatted catalog file.}$

Description

Prepare input file for helmsman from a helmsman formatted catalog file.

Usage

```
CreatehelmsmanOutput(
  catalog,
  out.dir = paste0(dirname(catalog), "/ExtrAttr/helmsman.results"),
  overwrite = FALSE
)
```

Arguments

catalog a catalog in ICAMS format. It can be a .csv file, or a matrix or data.frame.

Usually, it refers to "ground.truth.syn.catalog.csv".

out.dir Directory that will be created for the output; abort if it already exists. Usu-

ally, the out.dir will be a helmsman.results folder directly under the folder

storing catalog.

overwrite If TRUE, overwrite existing output

Details

Creates folder named helmsman.results containing catalogs in helmsman-formatted catalogs: Rows are signatures; the first column is the name of the mutation type, while the remaining columns are samples (tumors). These helmsman-formatted catalogs will the input when running helmsman program later on Python platform.

Value

 $invisible (\verb|catMatrix|), original catalog in helmsman format$

 ${\tt Create MultiModal MuSig Output}$

Prepare input file for MultiModalMuSig from a MultiModalMuSig formatted catalog file.

Description

Prepare input file for MultiModalMuSig from a MultiModalMuSig formatted catalog file.

Usage

```
CreateMultiModalMuSigOutput(
  catalog,
  out.dir = paste0(dirname(catalog), "/ExtrAttr/MultiModalMuSig.results"),
  overwrite = FALSE
)
```

Arguments

catalog a catalog in ICAMS format. It can be a .csv file, or a matrix or data.frame.

Usually, it refers to "ground.truth.syn.catalog.csv".

out.dir Directory that will be created for the output; abort if it already exists. Usu-

ally, the out.dir will be a MultiModalMuSig.results folder directly under

the folder storing catalog.

overwrite If TRUE, overwrite existing output

Details

Creates folder named MultiModalMuSig.results containing catalogs in MultiModalMuSig-formatted catalogs: Rows are signatures; the first column is the name of the mutation type, while the remaining columns are samples (tumors). These MM-formatted catalogs will the input when running MultiModalMuSig program later on Julia platform.

Value

invisible(catMatrix), original catalog in MultiModalMuSig format

helmsmanCatalog2ICAMS Read Catalog files or matrices in helmsman format.

Description

Read Catalog files or matrices in helmsman format.

Usage

```
helmsmanCatalog2ICAMS(
  cat,
  region = "unknown",
  catalog.type = "counts.signature"
)
```

Arguments

cat Input catalog, can be a tab-delimited text file in helmsman format, or a ma-

trix/data.frame object.

region Catalog region. Can be a specific genomic or exomic region, or "unknown".

Default: "unknown"

catalog.type Is the catalog a signature catalog, or a spectrum catalog? Default: "counts.signature"

Value

a catalog matrix in ICAMS format.

ICAMSCatalog2EMu

Convert Catalogs from ICAMS format to EMu format

Description

Convert Catalogs from ICAMS format to EMu format

Usage

ICAMSCatalog2EMu(catalog)

Arguments

catalog

A catalog matrix in ICAMS format. (SNS only!)

Value

a matrix without any dimnames, but the values are the transposition of the values in catalog.

ICAMSCatalog2helmsman Convert Catalogs from ICAMS format to helmsman format

Description

Convert Catalogs from ICAMS format to helmsman format

Usage

```
ICAMSCatalog2helmsman(catalog, type = "spectra")
```

Arguments

catalog

A catalog matrix in ICAMS format. (SNS only!)

type

Whether it is a spectra catalog ("spectra") or a signature catalog ("signature").

Value

a catalog matrix in helmsman format.

ICAMSCatalog2MM 7

MM format

ICAMSCatalog2MM	Convert	Catalogs	from	ICAMS	format to
ICAMSCALATORZMM	Converi	Calalogs	mom	ICAMS	jorniai io

Description

Convert Catalogs from ICAMS format to MM format

Usage

```
ICAMSCatalog2MM(catalog)
```

Arguments

catalog A catalog matrix in ICAMS format. (SNS/DNS/ID)

Value

a catalog matrix in MultiModalMuSig format.

MMCatalog2ICAMS Convert Catalogs (File or Matrix) from MM format to ICAMS format

Description

Convert Catalogs (File or Matrix) from MM format to ICAMS format

Usage

```
MMCatalog2ICAMS(cat, region = "unknown", catalog.type = "counts.signature")
```

Arguments

cat Input catalog, can be a tab-delimited file or matrix in MultiModalMuSig format.

region Catalog region. Can be a specific genomic or exomic region, or "unknown".

Default: "unknown"

catalog.type Is the catalog a signature catalog, or a spectrum catalog? Default: "counts.signature"

Value

a catalog matrix in ICAMS format.

PlotCatCOMPOSITE

Plot the a SignatureAnalyzer COMPOSITE signature or catalog into separate pdfs

Description

Plot the a Signature Analyzer COMPOSITE signature or catalog into separate pdfs

Usage

```
PlotCatCOMPOSITE(catalog, filename.header, type, id = colnames(catalog))
```

Arguments

catalog Catalog or signature matrix

filename.header

Contain path and the beginning part of the file name. The name of the pdf files will be: filename.header.SNS.96.pdf filename.header.SNS.1536.pdf

filename.header.DNS.78.pdf filename.header.ID.83.pdf

type See PlotCatalogToPdf.

id A vector containing the identifiers of the samples or signatures in catalog.

ReadAndAnalyzeExposures

Assess how well inferred exposures match input exposures We assume that in many cases attribution programs will be run outside of R on file inputs and will generate fill outputs.

Description

Assess how well inferred exposures match input exposures

We assume that in many cases attribution programs will be run outside of R on file inputs and will generate fill outputs.

```
ReadAndAnalyzeExposures(
  extracted.sigs,
  ground.truth.sigs,
  inferred.exp.path,
  ground.truth.exposures
)
```

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Arguments

extracted.sigs Path to file containing the extracted signature profiles.

ground.truth.sigs

File containing signature profiles from which the synthetic data were generated.

inferred.exp.path

File containing mutation counts (exposures) of synthetic tumors which are inferred to extracted or input signatures.

ground.truth.exposures

File containing the exposures from which the synthetic catalogs were generated. This file is used to restrict assessment of signature exposures to only those signatures in ground.truth.sigs that were actually represented in the exposures.

Details

Generates output files by calling TP_FP_FN_avg_sim

Value

A data.frame recording:

Ground.truth.exposure: sum of ground truth exposures of all tumors to all ground-truth signatures.

Inferred exposure: sum of inferred exposures of all tumors to all ground-truth signatures. Here, inferred exposure of a tumor to a ground-truth signature equals to the sum of the exposures of this tumor to all extracted signatures which are most similar to a ground-truth signature. If there is no extracted signature resembling an ground-truth signature, the inferred exposure of this ground-truth signature will be \emptyset .

Absolute.difference: sum of absolute difference between ground-truth exposure and inferred exposure of all tumors to all ground-truth signatures.

ReadAndAnalyzeSigs

Assess how well extracted signatures match input signatures We assume that in many cases extraction programs will be run outside of R on file inputs and will generate fill outputs.

Description

Assess how well extracted signatures match input signatures

We assume that in many cases extraction programs will be run outside of R on file inputs and will generate fill outputs.

Usage

ReadAndAnalyzeSigs(extracted.sigs, ground.truth.sigs, ground.truth.exposures)

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Arguments

```
extracted.sigs Path to file containing the extracted signature profiles.

ground.truth.sigs

File containing signature profiles from which the synthetic data were generated.

ground.truth.exposures
```

File containing the exposures from which the synthetic catalogs were generated. This file is used to restrict assessment to only those signatures in ground.truth.sigs that were actually represented in the exposures.

Details

Generates output files by calling TP_FP_FN_avg_sim

Value

```
See TP_FP_FN_avg_sim
```

ReadEMuCatalog

Read Catalog files in EMu format.

Description

Read Catalog files in EMu format.

Usage

```
ReadEMuCatalog(
  cat,
  mutTypes,
  sigOrSampleNames,
  region = "unknown",
  catalog.type = "counts.signature"
)
```

Arguments

cat A tab-delimited catalog text file in EMu format; or a EMu formatted matrix or

data.frame.

mutTypes Types of mutations. They are usually from an ICAMS:::catalog.row.header

object.

sigOrSampleNames

If input file is a counts signature file (catalog.type == "counts.signature"),

signature names should be provided.

If input file is a counts spectra file (catalog.type == "counts"), names of

samples should be provided.

region Catalog region. Can be a specific genomic or exomic region, or "unknown".

Default: "unknown"

catalog.type Is the catalog a signature catalog, or a spectrum catalog? Default: "counts"

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Value

a catalog matrix in ICAMS format.

ReadEMuExposureFile

Read Exposure files in EMu format.

Description

Read Exposure files in EMu format.

Usage

ReadEMuExposureFile(exposureFile, sigNames, sampleNames)

Arguments

exposureFile Exposure file generated by EMu. Usually, it is called "W_components.txt".

sigNames Names of signatures. These will be served as the rownames of the exposure

matrix.

sampleNames Names of samples in exposure file.

Return ICAMS/SynSigEval formatted exposure matrix.

ReadExposureMM

Read Catalog files in MM format

Description

Read Catalog files in MM format

Usage

ReadExposureMM(exposureFile)

Arguments

 ${\tt exposureFile} \qquad {\tt Input\ exposure\ file,\ can\ be\ a\ tab-delimited\ text\ file\ in\ MultiModalMuSig\ format.}$

Value

a exposure matrix in ICAMS format.

ReadhelmsmanExposure Read Exposure files in helmsman format.

Description

Read Exposure files in helmsman format.

Usage

ReadhelmsmanExposure(exposure, check.names = TRUE)

Arguments

exposure Exposure file generated by helmsman. Usually, it is called "W_components.txt".

check.names logical. If TRUE then the names of the variables in the data frame are checked

to ensure that they are syntactically valid variable names. If necessary they are adjusted (by make.names) so that they are, and also to ensure that there are no

duplicates.

Return ICAMS/SynSigEval formatted exposure matrix.

ReadSigProfilerExposure

Read a file containing exposures attributed by SigProfiler/Python

Description

Read a file containing exposures attributed by SigProfiler/Python

Usage

ReadSigProfilerExposure(file)

Arguments

file The name of the file to read.

Value

The corresponding signature matrix in standard internal representation.

RelabelExSigs 13

RelabelExSigs	Append most similar ground-truth signature and pairwise cosine similarity to the name of each extracted signature in matrix of extracted
	signatures.

Description

Append most similar ground-truth signature and pairwise cosine similarity to the name of each extracted signature in matrix of extracted signatures.

Usage

```
RelabelExSigs(sigAnalysis)
```

Arguments

sigAnalysis

A list returned by function ReadAndAnalyzeSigs, at least including:

- 1. ex.sigs: matrix of extracted signatures
- sim.matrix: full matrix between extracted signatures and ground-truth signatures
- 3. table: matrix include pairs of true positive ground-truth signatures and true positive extracted signatures.

Value

Matrix of extracted sigs, yet the names of signatures changed. New name: <old_name> (<name_of_most_similar_ground cosine_similarity) e.g., Sig.A -> Sig.A (SBS1 0.998)

SignatureAnalyzerSummarizeSBS1SBS5

Summarize all sub-directories of SignatureAnalyzer results on the correlated SBS1 / SBS5.

Description

This is special-purpose function to summarize results from one in-silico experiment that examines how well signatures can be extracted from synthetic tumors with correlated SBS1 and SBS5.

```
SignatureAnalyzerSummarizeSBS1SBS5(
  top.level.dir,
  summarize.exp = TRUE,
  overwrite = FALSE
)
```

Arguments

top.level.dir Path to top level directory.

summarize.exp Whether to summarize exposures when the file specified by inferred.exp.path exists.

overwrite If TRUE overwrite existing directories and files.

SignatureAnalyzerSummarizeTopLevel

Summarize all subdirectories of SignatureAnalyzer results on a major dataset.

Description

This function depends on a particular directory structure: see argument top.level.dir. This function finds the best of multiple SignatureAnalyzer extraction runs and summarizes the comparison of the best run with the ground truth.

Usage

```
SignatureAnalyzerSummarizeTopLevel(
  top.level.dir,
  summarize.exp = TRUE,
  overwrite = FALSE
)
```

Arguments

top.level.dir Path to top level directory, which must contain the following subdirectories:

- sa.sa.96/sa.results/
- sp.sp/sa.results/
- sa.sa.COMPOSITE/sa.results/
- sp.sa.COMPOSITE/sa.results/

Each of the directories must contain additional subdirectories, one for each SignatureAnalyzer run, names sa.run.<n>, where <n> is an integer (string of digits).

summarize.exp W

Whether to summarize exposures when the <run.dir>/<which.run>/sa.output.exp.csv exists.

overwrite

If TRUE overwrite and files in existing run.dir/summary folder.

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SplitCatCOMPOSITE	Split COMPOSITE (SNS1536+DBS78+ID83) catalogs in ICAMS for-
	mat into 3 individual catalogs.

Description

Split COMPOSITE (SNS1536+DBS78+ID83) catalogs in ICAMS format into 3 individual catalogs.

Usage

```
SplitCatCOMPOSITE(catalog)
```

Arguments

catalog Input catalog, can be a .csv file or matrix in ICAMS COMPOSITE format.

Value

a list, containing 3 catalog matrices in MultiModalMuSig format. Each matrix contains SNS1536, DBS78 and ID83 information, respectively.

SummarizeMultiRuns	Assess/evaluate multiple summarized runs on one dataset by one com-
	putational approach.

Description

Summarize results from each computational approach in resultPath/run.names (generated by running a computational approach), combine them into resultPath.

Usage

SummarizeMultiRuns(datasetName, toolName, resultPath, run.names)

 $by \ {\tt SummarizeSigOneExtrAttrSubdir}.$

Arguments

datasetName	Name of the dataset. (e.g. "S.0.1.Rsq.0.1"). Usually, it is has the same name as basename(top.dir).
toolName	Name of computational approach. (e.g. "SigProExtractor")
resultPath	Path expected to have multiple result folders each named as run.names (e.g. "seed.1"). The example resultPath is S.0.1.Rsq.0.1/sp.sp/ExtrAttr/hdp.results/ in old folder structure, or 3a.Original_output_K_unspecified/hdp/S.0.1.Rsq.0.1 in new folder structure.
run.names	A character vector records the list of directories which are under resultPath and contain results of computational approach, and a summary folder generated

Details

Also writes multiple files into folder resultPath.

Value

A list contain values of measures measures in multiple runs:

- \$averCosSim Average cosine similarity. Only similarities between TP sigs and extracted sigs most similar to them.
- \$truePos True Positives(TP): Ground-truth signatures which are active in the spectra, and extracted.
- \$falseNeg False Negatives(FN): Ground-truth signatures not extracted.
- \$falsePos False Positives(FP): Signatures wrongly extracted, not resembling any ground-truth signatures.
- \$TPR True positive rate (TPR, Sensitivity): TP / (TP + FN)
- \$PPV Positive predictive value (PPV, Precision): TP / (FP + TP)
- \$cosSim Cosine similarity between each of the ground-truth signatures, and its most similar extracted signature.
- \$AggManhattanDist (if exposures of signatures were inferred) Scaled Manhattan distance between ground-truth and inferred exposures to each of the ground-truth signatures.

This list also contains mean and sd, and other statistics of these measures in

- \$fivenum summary generated by fivenum columns of this table refer to Tukey's five number summary for each extraction measure across all runs:
 - min minimum
 - lower-hinge first quartile. Serve as the lower-hinge of the box-whisker plot.
 - median median of measure across all runs.
 - upper-hinge third quartile. Serve as the upper-hinge of the box-whisker plot.
 - max maximum
- \$fivenumMD Tukey's five number summary for aggregately-scaled Manhattan distance.
- \$meanSD mean and standard deviation for extraction measures.
- \$meanSDMD mean and standard deviation for aggregately-scaled Manhattan distance.

Summarize MultiTools MultiDatasets

Summarize results for multiple datasets, by different computational approaches.

Description

Summarize results of mutational signature extraction and exposure inference by multiple computational approaches on multiple datasets. Before running this function, make sure the summary file for each single data set toolSummaryPaths/OneToolSummary.Rda exists.

Usage

```
SummarizeMultiToolsMultiDatasets(
  toolSummaryPaths,
  out.dir,
  display.datasetName = FALSE,
  sort.by.composite.extraction.measure = "descending",
  overwrite = FALSE
)
```

Arguments

toolSummaryPaths

Paths of top-level dataset directories trees you want to investigate. E.g. "./S.0.1.Rsq.0.1" Note: OneToolSummary.RDa are expected to be exist under toolSummaryPaths.

out.dir Path of the output directory.

display.datasetName

Whether to put the name of spectra datasets inside of the csv outputs of summary tables.

sort.by.composite.extraction.measure

Whether to re-order the computational approaches on violin plots, based on the mean of composite measure.

"descending": Put the computational approach with the highest mean composite measure to the left, and arrange approaches in descending order. "ascending": Put the computational approach with the lowest mean composite measure to the left, and arrange approaches in ascending order. Anything else: Keep the computational approaches in a smart alphabetical order embedded with numbers, defined by mixedsort.

overwrite

Whether to overwrite the contents in out.dir if it already exists. (Default: FALSE)

Details

OneToolSummary.Rda is generated by SummarizeOneToolMultiDatasets).

SummarizeOneToolMultiDatasets

Combine results for multiple datasets, from one computational approaches.

Description

Summarize results from each computational approach in toolPath/datasetNames and combine them into out.dir.

```
SummarizeOneToolMultiDatasets(
  datasetNames = SynSigGen::SBS1SBS5datasetNames,
  datasetGroup,
  datasetGroupName,
  datasetSubGroup = NULL,
```

```
datasetSubGroupName = NULL,
toolName,
toolPath,
out.dir,
display.datasetName = FALSE,
overwrite = FALSE
)
```

Arguments

datasetNames

Names of datasets which are also folder names under toolPath. These folders contain results of toolName on such datasets. E.g. SynSigGen::SBS1SBS5datasetNames

datasetGroup

Numeric or character vector differentiating datasets within each group. E.g. For SBS1-SBS5 correlated datasets, we can consider the value of SBS1-SBS5 exposure ratio as the value for datsetgroup: rep(c(0.1,0.5,1,2,5,10),each

The value is set to Default if unspecified.

datasetGroupName

Meaning of all datasetGroup. E.g. For SBS1-SBS5 correlated datasets, we can consider "SBS1-SBS5 exposure ratio" as what datasetGroup is referring to.

datasetSubGroup

Numeric or character vector differentiating datasets within each sub-group. E.g. For SBS1-SBS5 correlated datasets, we can consider the value of SBS1-SBS5 correlation as the value of subgroup: rep(c(0.1,0.2,0.3,0.6),times = 5)

datasetSubGroupName

Meaning of all datasetSubGroup. E.g. For SBS1-SBS5 correlated datasets, we can consider "SBS1-SBS5 correlation" as what datasetSubGroup is referring to.

toolName

Name of computational approach to be investigated (e.g. "SigProExtractor")

toolPath

The path of the results of the computational approach to be investigated. May include top-level directory (e.g. 3a.Original_output_K_unspecified) and second-level directory containing outputs and summaries of one computational approach to be investigated (e.g. SigProExtractor or SigProExtractor.results).

One example: 3a.Original_output_K_unspecified/SigProExtractor

Note: this function expects file multiRun.RDa generated by SummarizeMultiRuns under toolPath/datasetNames

out.dir Path of the output directory.

display.datasetName

Whether to put the name of spectra datasets inside of the csv outputs of summary tables.

overwrite

Whether to overwrite the contents in out.dir if it already exists. (Default: FALSE)

 ${\tt SummarizeSigOneAttrSubdir}$

Assess/evaluate results from packages which can ONLY do exposure attribution.

Description

Packages including but not limited to: deconstructSigs, YAPSA.

Usage

```
SummarizeSigOneAttrSubdir(
  run.dir,
  ground.truth.exposure.dir = paste0(run.dir, "/../../"),
  overwrite = FALSE,
  summary.folder.name = "summary",
  export.Manhattan.each.spectrum = FALSE
)
```

Arguments

run.dir

Lowest level path to results, e.g. <top.dir>/sa.sa.96/Attr/YAPSA.results/seed.1/ Here, <top.dir> refers to a top-level directory which contains the full information of a synthetic dataset. (e.g. syn.2.7a.7b.abst.v8) This code depends on a conventional directory structure documented elsewhere. For packages which can do both extraction and attribution, we expect two files, ground.truth.signatures.csv and inferred.exposures.csv are in the folder.

ground.truth.exposure.dir

Folder which stores ground-truth exposures. It defaults to be sub.dir, i.e. run.dir/../../

overwrite

If TRUE overwrite existing directories and files.

summary.folder.name

The name of the folder containing summary results. Usually, it equals to "summary".

 ${\tt export.Manhattan.each.spectrum}$

Whether to export csv files for Manhattan distance of each mutational spectrum.

Details

Here, we excluded SignatureEstimation. Although it is also a package with only attribution, but it has two attribution algorithms. Therefore the naming of the results are slightly different from the other two packages.

 ${\tt SummarizeSigOneExtrAttrSubdir}$

Assess/evaluate results from packages which can do BOTH extraction and attribution, excluding SigProfiler-Python and SignatureAnalyzer.

Description

Packages including but not limited to: hdp, MutationalPatterns, sigfit, signeR, SomaticSignatures.

Usage

```
SummarizeSigOneExtrAttrSubdir(
  run.dir,
  ground.truth.exposure.dir = paste0(run.dir, "/../../"),
  summarize.exp = TRUE,
  overwrite = FALSE,
  summary.folder.name = "summary",
  export.Manhattan.each.spectrum = FALSE
)
```

Arguments

run.dir A directory which contains output of computational approach in one run on a

specific dataset, possibly with a specified seed. E.g. 2b.Full_output_K_as_2/hdp.results/S.0.1

This code depends on a conventional directory structure documented in NEWS.md.

ground.truth.exposure.dir

Folder which stores ground-truth exposures. Should contain a file named ground. truth. syn. exposure PCAWG paper: run.dir/../../ In SBS1-SBS5 paper: 0.Input_datasets/S.0.1.Rsq.0.1/

summarize.exp Whether to summarize exposures when the file specified by inferred.exp.path

exists.

overwrite If TRUE overwrite and files in existing run.dir/summary folder.

summary.folder.name

The name of the folder containing summary results. Usually, it equals to "summary".

export.Manhattan.each.spectrum

Whether to export csv files for Manhattan distance of each mutational spectrum.

 ${\tt SummarizeSigOnehelmsmanSubdir}$

Assess/evaluate results from helmsman.NMF

Description

Assess/evaluate results from helmsman.NMF

```
SummarizeSigOnehelmsmanSubdir(
  run.dir,
  ground.truth.exposure.dir = paste0(run.dir, "/../../"),
  summarize.exp = TRUE,
  overwrite = FALSE
)
```

Arguments

run.dir A directory which contains output of helmsman.NMF in one run on a specific

dataset, possibly with a specified seed. E.g. 2b.Full_output_K_as_2/helmsman.NMF.results/S.@

This code depends on a conventional directory structure documented in NEWS.md.

ground.truth.exposure.dir

 $Folder\ which\ stores\ ground-truth\ exposures.\ Should\ contain\ a\ file\ named\ ground\ .\ truth\ .\ syn\ .\ exposures\ .$

In PCAWG paper: run.dir/../../ In SBS1-SBS5 paper: 0.Input_datasets/S.0.1.Rsq.0.1/ Whether to summarize exposures when the file specified by inferred.exp.path

summarize.exp Wheth exists.

overwrite If TRUE overwrite and files in existing run.dir/summary folder.

 ${\tt SummarizeSigOneSigProExtractorSubdir}$

Assess/evaluate results from SigProExtractor (v0.0.5.45+)

Description

SigProfiler-python de novo extraction and attribution package. Assessment is restricted to $v0.0.5.43 \sim v0.0.5.77$, because different version has different folder structure.

Usage

```
SummarizeSigOneSigProExtractorSubdir(
  run.dir,
  ground.truth.exposure.dir = paste0(run.dir, "/../../"),
  summarize.exp = TRUE,
  overwrite = FALSE,
  hierarchy = FALSE,
  summary.folder.name = "summary",
  export.Manhattan.each.spectrum = FALSE
)
```

Arguments

run.dir A directory which contains output of SigProExtractor in one run on a specific

dataset, possibly with a specified seed. E.g. 2b.Full_output_K_as_2/SigProExtractor.results/

This code depends on a conventional directory structure documented in NEWS.md.

ground.truth.exposure.dir

Folder which stores ground-truth exposures. Should contain a file named ground. truth. syn. exposures.

 $In\ PCAWG\ paper:\ run.dir/../../\ In\ SBS1-SBS5\ paper:\ 0.\ Input_datasets/S.0.1.\ Rsq.\ 0.1/datasets/S.0.1.$

 $summarize.exp \qquad Whether to summarize exposures when the file specified by \verb"inferred.exp.path" and exp. path an$

exists.

overwrite If TRUE overwrite and files in existing run.dir/summary folder.

hierarchy Whether the user have enabled hierarchy = True when running SigProExtractor.

specifying True or False into SigProExtractor will cause the program to generate

different folder structure.

summary.folder.name

The name of the folder containing summary results. Usually, it equals to "sum-

mary".

export.Manhattan.each.spectrum

Whether to export csv files for Manhattan distance of each mutational spectrum.

Details

This function cannot be used on new SigProfilerExtractor (v1+) as the folder structure has been changed markedly

 ${\tt SummarizeSigOneSigProSSSubdir}$

Assess/evaluate results from sigproSS (a.k.a. SigProfiler Python attribution package)

Description

Assess/evaluate results from sigproSS (a.k.a. SigProfiler Python attribution package)

Usage

```
SummarizeSigOneSigProSSSubdir(
  run.dir,
  ground.truth.exposure.dir = paste0(run.dir, "/../../"),
  overwrite = FALSE,
  summary.folder.name = "summary",
  export.Manhattan.each.spectrum = FALSE
)
```

Arguments

run.dir

Lowest level path to results, e.g. <top.dir>/sa.sa.96/ExtrAttr/SigProExtractor.results/see Here, <top.dir> refers to a top-level directory which contains the full information of a synthetic dataset. (e.g. syn.2.7a.7b.abst.v8) This code depends on a conventional directory structure documented elsewhere. However there should be a directory <run.dir>/SBS96 which stores SigProfiler results.

ground.truth.exposure.dir

TODO(Wu Yang): Fix this File name which stores ground-truth exposures; defaults to "ground.truth.syn.exposures.csv". This file can be found in the sub.dir, i.e. <run.dir>/../../

overwrite If TRUE overwrite existing directories and files.

summary.folder.name

The name of the folder containing summary results. Usually, it equals to "summary".

export.Manhattan.each.spectrum

Whether to export csv files for Manhattan distance of each mutational spectrum.

 ${\tt SummarizeSigProExtractor}$

Summarize SigProfiler results in the sa.sa.96 and/or sp.sp subdirectories.

Description

Summarize SigProfiler results in the sa.sa.96 and/or sp.sp subdirectories.

Usage

```
SummarizeSigProExtractor(
  top.dir,
  sub.dir = c("sa.sa.96", "sp.sp"),
  overwrite = FALSE
)
```

Arguments

top.dir	The top directory of a conventional data structure containing at least one of the subdirectories: sa.sa.96/sp.results and sp.sp/sp.results; see further documentation elsewhere.
sub.dir	The subdirectory under top.dir, and containing a folder named sp.results. By default, it contains both c("sa.sa", "sp.sp"). But you should specify sub.dir = "sp.sp" for top.dir with only the sp.sp subdirectory (as is the case for the correlated SBS1-and-SBS5-containing data sets).
overwrite	If TRUE overwrite and files in existing run.dir/summary folder.

Details

Results are put in standardized subdirectories of top. $\mbox{\tt dir}.$

Description

Assess the performance of two steps in mutational signature analysis:

- · signature extraction
- exposure inference (a.k.a. signature attribution)

by computational approaches, using catalogs of synthetic mutational spectra created by package SynSigGen.

24 SynSigEval

Input

SynSigEval requires the input data listed below:

- 1. E, matrix of synthetic exposures (signatures x samples)
- 2. S, mutational signature profiles (mutation type x signature)
- 3. synthetic.spectra, synthetic mutational spectra with known ground-truth mutational signature profiles (S) and exposures (synthetic.exposures). It can be created from SynSigGen.
- 4. T, signatures extracted by SignatureAnalyzer, SigProfiler, or other computational approaches on synthetic.spectra. For attribution-only approaches, T=S.
- 5. F, exposures inferred by computational approaches on synthetic.spectra.

Folder structure for SynSigEval v0.2

Summary function will fit to the new 5-level folder structure:

First Level - top.level.dir: dataset folder (e.g. "S.0.1.Rsq.0.1", "syn.pancreas"). All spectra datasets under any top.level.dir have the same exposure.

Second Level - ground.truth.exposure.dir: spectra folder: (e.g. "sp.sp", "sa.sa.96"). All spectra datasets under any second.level.dir have the same signature and the same exposure counts.

Third Level - third.level.dir: It can be ("Attr") for storing results of packages which can only do exposure attribution of known signatures ("Attr"); it can also be ("ExtrAttr"), folder to store results of software packages which can do de-novo extraction and following attribution.

Fourth Level - tool.dir: The results of a software package (e.g. "SigProExtractor.results", "SignatureEstimation.QP.resu Under this level, tool.dir may contain multiple run.dir, each is a run of the software package using a specific number of seed.

Fifth level - run.dir: contains results from a run of the software package using a specific number of seed. (e.g. "seed.1")

Summarize results

1. Summarize results in fifth-level run.dir:

Relevant functions are:

- SummarizeSigProExtractor
- SignatureAnalyzerSummarizeTopLevel
- SignatureAnalyzerSummarizeSBS1SBS5
- SummarizeSigOneExtrAttrSubdir
- SummarizeSigOneAttrSubdir
- SummarizeSigOnehelmsmanSubdir
- $\bullet \ {\tt SummarizeSigOneSigProSSSubdir}$
- 2. Summarize results of multiple runs by a computational approach on one spectra data set: SummarizeMultiRuns
- 3. Summarize results of multiple computational approaches on one spectra data set: SummarizeMultiToolsOneDataset
- 4. Summarize results of multiple computational approaches on multiple spectra data sets: SummarizeMultiToolsMultiDatasets

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