Package 'SynSigEval'

December 24, 2020

```
Type Package
Title Evaluate Results of Mutational Signature Analysis Software
     on Synthetic Spectra Created by Package SynSigGen
Version 0.1.8
Author Steven G. Rozen, Yang Wu
Maintainer Steven G. Rozen <steverozen@gmail.com>
     Examine and evaluate the output of mutational signature analysis computational approaches.
License GPL-3
Language en-US
Encoding UTF-8
LazyData true
Imports data.table,
     ICAMS,
     ICAMSxtra (>= 0.0.2),
     stats,
     devtools,
     utils,
     rlang,
     graphics,
     grDevices,
     ggplot2,
     ggbeeswarm,
     gtools
Remotes github::steverozen/ICAMSxtra@*release
Depends R (>= 3.5)
RoxygenNote 7.1.1
Suggests testthat,
     knitr,
     lsa,
     rmarkdown,
     DelayedArray,
     BiocManager,
     ggpubr
```

R topics documented:

	CopyBestSignatureAnalyzerResult	2
	CreateEMuOutput	3
	CreatehelmsmanOutput	4
	CreateMultiModalMuSigOutput	4
	helmsmanCatalog2ICAMS	5
	ICAMSCatalog2EMu	6
	ICAMSCatalog2helmsman	6
	ICAMSCatalog2MM	7
	MMCatalog2ICAMS	7
	PlotCatCOMPOSITE	8
	ReadAndAnalyzeExposures	8
	ReadAndAnalyzeSigs	9
	ReadEMuCatalog	10
	ReadEMuExposureFile	11
	ReadExposureMM	11
	ReadhelmsmanExposure	12
	ReadSigProfilerExposure	12
	ReadSigProfilerSigDBS78	13
	ReadSigProfilerSigSBS96	13
	SignatureAnalyzerSummarizeSBS1SBS5	14
		14
	SplitCatCOMPOSITE	15
	SummarizeMultiRuns	15
	SummarizeMultiToolsMultiDatasets	16
	SummarizeMultiToolsOneDataset	17
	SummarizeOneToolMultiDatasets	18
	SummarizeSigOneAttrSubdir	19
	SummarizeSigOneExtrAttrSubdir	20
	SummarizeSigOnehelmsmanSubdir	21
	SummarizeSigOneSigProExtractorSubdir	21
	SummarizeSigOneSigProSSSubdir	22
	SummarizeSigProExtractor	
	SynSigEval	
Index		25

 ${\tt CopyBestSignatureAnalyzerResult}$

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/

CreateEMuOutput 3

Usage

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

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

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

 ${\tt Create Multi Modal MuSig Output}$

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.

Usage

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

ReadAndAnalyzeSigs 9

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 MatchSigsAndRelabel

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)

10 ReadEMuCatalog

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 MatchSigsAndRelabel

Value

See MatchSigsAndRelabel

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"

ReadEMuExposureFile 11

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.

ReadSigProfilerSigDBS78

Read a file containing DBS78 signatures extracted by SigProfiler/Python

Description

Read a file containing DBS78 signatures extracted by SigProfiler/Python

Usage

```
ReadSigProfilerSigDBS78(file)
```

Arguments

file

The name of the file to read.

Value

The corresponding signature matrix in standard internal representation.

ReadSigProfilerSigSBS96

Read a file containing SBS96 signatures extracted by SigPro-filer/Python

Description

Read a file containing SBS96 signatures extracted by SigProfiler/Python

Usage

```
ReadSigProfilerSigSBS96(file)
```

Arguments

file

The name of the file to read.

Value

The corresponding signature matrix in standard internal representation.

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.

Usage

SignatureAnalyzerSummarizeSBS1SBS5(top.level.dir, overwrite = FALSE)

Arguments

```
top.level.dir Path to top level directory.
```

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, 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).

overwrite If TRUE overwrite existing summary files.

15 **SplitCatCOMPOSITE**

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

Description

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

Usage

SplitCatCOMPOSITE(catalog)

Arguments

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

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 for one dataset from one
	computational approach.

Description

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

Usage

SummarizeMultiRuns(datasetName, toolName, tool.dir, run.names)

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")
tool.dir	Fourth level path from the top.dir. Expected to have multiple runs with different names (e.g. "seed.1") That is, top.dir/sp.sp/ExtrAttr/sa.results/. or top.dir/sa.sa.96/Attr/deconstructSigs.results/
	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 within the tool.dir which stores the software output.
run.names	A character vector records the list of run.dir, or fifth level directories from the

dataset top-level folder. E.g., c("seed.1", "seed.691")

Details

Also writes multiple files into folder tool.dir.

Value

A list contain values of measures measures in multiple runs:

- \$averCosSim Cosine similarity
- \$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): TP / (FP + TP)
- \$cosSim Average cosine similarity to each of the ground-truth signatures.
- \$AggManhattanDist 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
- \$fivenumMD
- \$meanSD
- \$meanSDMD

SummarizeMultiToolsMultiDatasets

Summarize results for multiple datasets, by different computational approaches.

Description

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

Usage

```
SummarizeMultiToolsMultiDatasets(
  dataset.dirs,
  second.third.level.dirname,
  out.dir,
  overwrite = FALSE
)
```

Arguments

```
dataset.dirs
                  Paths of top-level dataset directories trees you want to investigate. E.g. "./S.0.1.Rsq.0.1"
second.third.level.dirname
```

Name of the second.level.dir (e.g. "sp.sp") and the third.level.dir (e.g. "Ex-

trAttr") to be investigated.

Examples are: "sp.sp/ExtrAttr", "sa.sa.96/Attr"

Note: multiTools.RDa are expected to be exist under dataset.dirs/second.third.level.dirnam

out.dir Path of the output directory.

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

Details

multiTools.Rda is generated by SummarizeMultiToolsOneDataset).

SummarizeMultiToolsOneDataset

Combine results for a single dataset, from different computational approaches.

Description

Summarize results from each computational approach in third.level.dir/tool.dirnames (generated by SummarizeMultiRuns), combine them into third.level.dir.

Usage

```
SummarizeMultiToolsOneDataset(
  third.level.dir,
  toolNames,
  tool.dirnames,
  datasetGroup,
  datasetGroupName,
  datasetSubGroup = NULL,
  datasetSubGroupName = NULL
)
```

Arguments

third.level.dir

Third level path distinguishing de novo extraction + attribution packages from attribution-only packages. Examples: top.dir/sp.sp/ExtrAttr/top.dir/sa.sa/Attr/

toolNames Names of computational approach. (e.g. "SigProExtractor")

tool.dirnames

Third level path from the top.dir. Expected to have summarized results generated by SummarizeMultiRuns. (multiRun.RDa, ManhattanDist.csv, meanSD.csv, meanSD.Manhattan.dist.csv) Examples: "signeR.results" (Under third.level.dir "ExtrAttr") "deconstructSigs.results" (Under third.level.dir "Attr") 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 within the tool. names which stores the software output.

datasetGroup

Numeric or character vector specifying the groups each dataset belong to. E.g.

For SBS1-SBS5 correlated datasets, we can consider slope as the group: c("slope=0.1","slope=0.5","s Default: "Default"

datasetGroupName

Meaning or label of all datasetGroup. E.g. For SBS1-SBS5 correlated datasets, we can consider "SBS1:SBS5 mutation count ratio" as the label of the datasetGroup slope.

datasetSubGroup

Optional. Numeric or character vector differentiating datasets within each group. E.g. For SBS1-SBS5 correlated datasets, we can consider Pearson's R^2 as the subgroup: c("Rsq=0.1","Rsq=0.2","Rsq=0.3","Rsq=0.6") Default: Names of datasets, which are basename(dataset.dirs)

datasetSubGroupName

Optional. Meaning or label of all datasetSubGroup. E.g. For SBS1-SBS5 correlated datasets, we can consider "Pearson's R squared" as the label of the datasetSubGroup Pearson's R^2.

Details

This function generates multiTools.RDa under third.level.dir

Value

A list contain c(mean,sd) of multiple runs: Cosine similarity True Positives(TP): Ground-truth signatures which are active in the spectra, and extracted. False Negatives(FN): Ground-truth signatures not extracted. False Positives(FP): Signatures wrongly extracted, not resembling any ground-truth signatures. True positive rate (TPR, Sensitivity): TP / (TP + FN) Positive predictive value (PPV, Precision): TP / (FP + TP)

 ${\tt SummarizeOneToolMultiDatasets}$

Combine results for multiple datasets, from one computational approaches.

Description

Summarize results from each computational approach in third.level.dir/tool.dirnames (generated by SummarizeMultiRuns), combine them into third.level.dir.

Usage

```
SummarizeOneToolMultiDatasets(
  dataset.dirs,
  datasetGroup,
  datasetGroupName,
  datasetSubGroup = NULL,
  datasetSubGroupName = NULL,
  toolName,
  tool.dirname,
  out.dir,
  overwrite = FALSE
)
```

Arguments

dataset.dirs Paths of top-level dataset directories trees you want to investigate. E.g. "./S.0.1.Rsq.0.1"

datasetGroup Numeric or character vector specifying the group each dataset belong to. E.g.

For SBS1-SBS5 correlated datasets, we can consider slope (SBS1:SBS5 count

ratio) as the group: c(0.1,0.5,1,2,5,10) Default: "Default"

datasetGroupName

Meaning or label of all datasetGroup. E.g. For SBS1-SBS5 correlated datasets, we can consider "SBS1:SBS5 mutation count ratio" as the label of the datasetGroup slope.

datasetSubGroup

Numeric or character vector differentiating datasets within each group. E.g. For SBS1-SBS5 correlated datasets, we can consider Pearson's R^2 as the subgroup: c(0.1,0.2,0.3,0.6) Default: Names of datasets, which are basename(dataset.dirs)

datasetSubGroupName

Meaning or label of all datasetSubGroup. E.g. For SBS1-SBS5 correlated datasets, we can consider "Pearson's R squared" as the label of the datasetSubGroup

Pearson's R^2.

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

tool.dirname Name of the second.level.dir (e.g. "sp.sp"), third.level.dir (e.g. "ExtrAttr") and

 $tool.dir\ (e.g.\ "SigProExtractor.results")\ to\ be\ investigated.$

 $One\ example:\ "sp.sp/ExtrAttr/SigProExtractor.results"$

Note: this function expects the summary generated by SummarizeSigOneSubdir

under dataset.dirs/tool.dirname

out.dir Path of the output directory.

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
)
```

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.

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, "/../../"),
  overwrite = FALSE
)
```

Arguments

run.dir

Lowest level path to result of a run. E.g. <top.dir>/sa.sa.96/ExtrAttr/SomaticSignatures.res 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, extracted.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.

 ${\tt SummarizeSigOnehelmsmanSubdir}$

Assess/evaluate results from SigProfiler-python (a.k.a. SigProExtractor) Assessment is restricted to v0.0.5.43, because different version has different folder structure.

Description

Assess/evaluate results from SigProfiler-python (a.k.a. SigProExtractor) Assessment is restricted to v0.0.5.43, because different version has different folder structure.

Usage

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

Folder which stores ground-truth exposures. Usually, it refers to sub.dir, i.e.

run.dir/../../

overwrite

If TRUE overwrite existing directories and files.

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. (Default: FALSE)

 ${\tt SummarizeSigOneSigProExtractorSubdir}$

Assess/evaluate results from SigProExtractor SigProfiler-python de novo extraction and attribution package. Assessment is restricted to v0.0.5.43+, because different version has different folder structure.

Description

Assess/evaluate results from SigProExtractor SigProfiler-python de novo extraction and attribution package. Assessment is restricted to v0.0.5.43+, because different version has different folder structure.

Usage

```
SummarizeSigOneSigProExtractorSubdir(
  run.dir,
  ground.truth.exposure.dir = paste0(run.dir, "/../../"),
  overwrite = FALSE,
  hierarchy = 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.

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. (Default: FALSE)

 ${\tt SummarizeSigOneSigProSSSubdir}$

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
)
```

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.

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	whether to overwrite the existing run.dir/summary folder? If chosen to be FALSE and there is an existing summary folder, an error will be raised.

Details

Results are put in standardized subdirectories of top.dir.

SynSigEval	SynSigEval
SynSigEval	SynsigEvai

Description

Assess the performance of two steps in mutational signature analysis:

- signature extraction
- exposure inferrence (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 SignatureAnalzer, 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

Index

```
BestSignatureAnalyzerResult, 3
CopyBestSignatureAnalyzerResult, 2
CreateEMuOutput, 3
CreatehelmsmanOutput, 4
                                                SynSigEval, 23
CreateMultiModalMuSigOutput, 4
data.frame, 9
\verb|helmsmanCatalog2ICAMS|, 5
ICAMSCatalog2EMu, 6
ICAMSCatalog2helmsman, 6
ICAMSCatalog2MM, 7
make.names, 12
MatchSigsAndRelabel, 9, 10
MMCatalog2ICAMS, 7
PlotCatalogToPdf, 8
PlotCatCOMPOSITE, 8
ReadAndAnalyzeExposures, 8
ReadAndAnalyzeSigs, 9
ReadEMuCatalog, 10
ReadEMuExposureFile, 11
ReadExposureMM, 11
ReadhelmsmanExposure, 12
ReadSigProfilerExposure, 12
ReadSigProfilerSigDBS78, 13
ReadSigProfilerSigSBS96, 13
SignatureAnalyzerSummarizeSBS1SBS5, 14,
SignatureAnalyzerSummarizeTopLevel, 14,
        24
SplitCatCOMPOSITE, 15
SummarizeMultiRuns, 15, 17, 18
{\tt Summarize MultiTools MultiDatasets}, 16
SummarizeMultiToolsOneDataset, 17, 17
SummarizeOneToolMultiDatasets, 18
SummarizeSigOneAttrSubdir, 19, 24
SummarizeSigOneExtrAttrSubdir, 20, 24
```

SummarizeSigOnehelmsmanSubdir, 21, 24

SummarizeSigOneSigProExtractorSubdir, 21
SummarizeSigOneSigProSSSubdir, 22, 24
SummarizeSigProExtractor, 23, 24
SynSigEval, 23