

Package ‘SynSigEval’

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Type Package

Title Evaluate Results of Mutational Signature Analysis Software
on Synthetic Spectra Created by Package SynSigGen

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Author Steven G. Rozen, Yang Wu

Maintainer Steven G. Rozen <steverozen@gmail.com>

Description

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

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Imports fs,
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grDevices,
ggplot2,
ggpubr,
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ICAMS (>= 3.0.5),
mSigTools (>= 1.0.2),
methods,
rlang,
stats,
utils

Remotes github::steverozen/ICAMS,
github::Rozen-Lab/mSigTools

Depends R (>= 3.5)

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Suggests BiocManager,
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rmarkdown,
testthat

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	10
ReadExposureMM	11
ReadhelmsmanExposure	11
ReadSigProfilerExposure	12
RelabelExSigs	12
SignatureAnalyzerSummarizeSBS1SBS5	13
SignatureAnalyzerSummarizeTopLevel	13
SplitCatCOMPOSITE	14
SummarizeMultiRuns	14
SummarizeMultiToolsMultiDatasets	16
SummarizeOneToolMultiDatasets	17
SummarizeSigOneAttrSubdir	18
SummarizeSigOneExtrAttrSubdir	19
SummarizeSigOnehelmsmanSubdir	20
SummarizeSigOneSigProExtractorSubdir	20
SummarizeSigOneSigProSSSubdir	21
SummarizeSigProExtractor	22
SynSigEval	23
Index	25

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/

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	<i>Prepare input file for EMu from a EMu formatted catalog file.</i>
-----------------	--

Description

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

Usage

```
CreateEMuOutput(
  catalog,
  out.dir = paste0(dirname(catalog), "/ExtraAttr/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. Usually, 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 be the input when running EMu program later on compiled binary.

Value

invisible(catalog), original catalog in EMu format

CreatehelmsmanOutput	<i>Prepare input file for helmsman from a helmsman formatted catalog file.</i>
----------------------	--

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. Usually, 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 be the input when running helmsman program later on Python platform.

Value

`invisible(catMatrix)`, original catalog in helmsman format

CreateMultiModalMuSigOutput	<i>Prepare input file for MultiModalMuSig from a MultiModalMuSig formatted catalog file.</i>
-----------------------------	--

Description

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

Usage

```
CreateMultiModalMuSigOutput(
  catalog,
  out.dir = paste0(dirname(catalog), "/ExtraAttr/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. Usually, 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 be 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 matrix/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	<i>Convert Catalogs from ICAMS format to MM format</i>
-----------------	--

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	<i>Convert Catalogs (File or Matrix) from MM format to ICAMS format</i>
-----------------	---

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	<i>Plot the a SignatureAnalyzer COMPOSITE signature or catalog into separate pdfs</i>
------------------	---

Description

Plot the a SignatureAnalyzer 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

Description

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,
  reference.sigs,
  inferred.exp.path,
  ground.truth.exposures
)
```

Arguments

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

Inferred.exposure: sum of inferred exposures of all tumors to all reference 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 reference signature. If there is no extracted signature resembling an ground-truth signature, the inferred exposure of this ground-truth signature will be 0.

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

ReadAndAnalyzeSigs	<i>Assess how well extracted signatures match input signatures</i>
--------------------	--

Description

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, reference.sigs, ground.truth.exposures)
```

Arguments

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

`reference.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 `reference.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	<i>Read Catalog files in EMu format.</i>
----------------	--

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	<p>If input file is a counts signature file (catalog.type == "counts.signature"), signature names should be provided.</p> <p>If input file is a counts spectra file (catalog.type == "counts"), names of samples should be provided.</p>
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"

Value

a catalog matrix in ICAMS format.

ReadEMuExposureFile	<i>Read Exposure files in EMu format.</i>
---------------------	---

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

exposureFile	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

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

Description

Append most similar reference 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
2. sim.matrix: full matrix between extracted signatures and reference signatures
3. table: matrix include pairs of true positive reference 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_truth> 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.

Usage

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

Arguments

<code>top.level.dir</code>	Path to top level directory.
<code>summarize.exp</code>	Whether to summarize exposures when the file specified by <code>inferred.exp.path</code> exists.
<code>overwrite</code>	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: <ul style="list-style-type: none"> • 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	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.

SplitCatCOMPOSITE	<i>Split COMPOSITE (SNS1536+DBS78+ID83) catalogs in ICAMS format into 3 individual catalogs.</i>
-------------------	--

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	<i>Assess/evaluate multiple summarized runs on one dataset by one computational approach.</i>
--------------------	---

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)
```

Arguments

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

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

SummarizeMultiToolsMultiDatasets

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

<code>toolSummaryPaths</code>	Paths of top-level dataset directories trees you want to investigate. E.g. <code>"/S.0.1.Rsq.0.1"</code> Note: <code>OneToolSummary.Rda</code> are expected to be exist under <code>toolSummaryPaths</code> .
<code>out.dir</code>	Path of the output directory.
<code>display.datasetName</code>	Whether to put the name of spectra datasets inside of the csv outputs of summary tables.
<code>sort.by.composite.extraction.measure</code>	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 .
<code>overwrite</code>	Whether to overwrite the contents in <code>out.dir</code> 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`.

Usage

```
SummarizeOneToolMultiDatasets(
  datasetNames = SynSigGen::SBS1SBS5datasetNames,
  datasetGroup,
  datasetGroupName,
  datasetSubGroup = NULL,
  datasetSubGroupName = NULL,
  toolName,
  toolPath,
  out.dir,
  display.datasetName = FALSE,
  overwrite = FALSE
)
```

Arguments

- | | |
|----------------------------------|--|
| <code>datasetNames</code> | Names of datasets which are also folder names under <code>toolPath</code> . These folders contain results of <code>toolName</code> on such datasets. E.g. <code>SynSigGen::SBS1SBS5datasetNames</code> |
| <code>datasetGroup</code> | 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 <code>datasetGroup</code> : <code>rep(c(0.1, 0.5, 1, 2, 5, 10), each = 4)</code>
The value is set to Default if unspecified. |
| <code>datasetGroupName</code> | Meaning of all <code>datasetGroup</code> . E.g. For SBS1-SBS5 correlated datasets, we can consider "SBS1-SBS5 exposure ratio" as what <code>datasetGroup</code> is referring to. |
| <code>datasetSubGroup</code> | 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 <code>subgroup</code> : <code>rep(c(0.1, 0.2, 0.3, 0.6), times = 5)</code> |
| <code>datasetSubGroupName</code> | Meaning of all <code>datasetSubGroup</code> . E.g. For SBS1-SBS5 correlated datasets, we can consider "SBS1-SBS5 correlation" as what <code>datasetSubGroup</code> is referring to. |
| <code>toolName</code> | 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)

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, reference.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".
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.

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 In 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.

SummarizeSigOnehelmsmanSubdir

Assess/evaluate results from helmsman.NMF

Description

Assess/evaluate results from helmsman.NMF

Usage

```
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.0.1.1. 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. In 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.

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 ~ 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

<code>run.dir</code>	A directory which contains output of SigProExtractor in one run on a specific dataset, possibly with a specified seed. E.g. <code>2b.Full_output_K_as_2/SigProExtractor.results/</code> This code depends on a conventional directory structure documented in NEWS.md.
<code>ground.truth.exposure.dir</code>	Folder which stores ground-truth exposures. Should contain a file named <code>ground.truth.syn.exposure</code> In PCAWG paper: <code>run.dir/../../../../</code> In SBS1-SBS5 paper: <code>0.Input_datasets/S.0.1.Rsq.0.1/</code>
<code>summarize.exp</code>	Whether to summarize exposures when the file specified by <code>inferred.exp.path</code> exists.
<code>overwrite</code>	If TRUE overwrite and files in existing <code>run.dir/summary</code> folder.
<code>hierarchy</code>	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.
<code>summary.folder.name</code>	The name of the folder containing summary results. Usually, it equals to "summary".
<code>export.Manhattan.each.spectrum</code>	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

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

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

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.dir.

SynSigEval	<i>SynSigEval</i>
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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.

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.results"). 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](#)
 - [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](#)

CreateMultiModalMuSigOutput, [4](#)

data.frame, [9](#)

fivenum, [15](#)

helmsmanCatalog2ICAMS, [5](#)

ICAMSCatalog2EMu, [6](#)

ICAMSCatalog2helmsman, [6](#)

ICAMSCatalog2MM, [7](#)

make.names, [11](#)

mixedsort, [16](#)

MMCatalog2ICAMS, [7](#)

PlotCatalogToPdf, [8](#)

PlotCatCOMPOSITE, [8](#)

ReadAndAnalyzeExposures, [8](#)

ReadAndAnalyzeSigs, [9](#), [12](#)

ReadEMuCatalog, [10](#)

ReadEMuExposureFile, [10](#)

ReadExposureMM, [11](#)

ReadhelmsmanExposure, [11](#)

ReadSigProfilerExposure, [12](#)

RelabelExSigs, [12](#)

SignatureAnalyzerSummarizeSBS1SBS5, [13](#),
[23](#)

SignatureAnalyzerSummarizeTopLevel, [13](#),
[23](#)

SplitCatCOMPOSITE, [14](#)

SummarizeMultiRuns, [14](#)

SummarizeMultiToolsMultiDatasets, [16](#)

SummarizeOneToolMultiDatasets, [16](#), [17](#)

SummarizeSigOneAttrSubdir, [18](#), [24](#)

SummarizeSigOneExtrAttrSubdir, [15](#), [19](#),
[24](#)

SummarizeSigOnehelmsmanSubdir, [20](#), [24](#)

SummarizeSigOneSigProExtractorSubdir,
[20](#)

SummarizeSigOneSigProSSSubdir, [21](#), [24](#)

SummarizeSigProExtractor, [22](#), [23](#)

SynSigEval, [23](#)

TP_FP_FN_avg_sim, [9](#)