# Package 'sampleContamination'

February 8, 2018

| Title Sample Contamination R Package                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |   |
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| <b>Description</b> Sample contamination discovery using variant allele frequency (VAF)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |   |
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| License Roswell Park Cancer Institute                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |   |
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2 filterCommonSNPs

filterByCov

Filter mutations base on coverage

## **Description**

This function filters mutation base on coverage

## Usage

```
filterByCov(VAFdata, VAFcov, minCov)
```

## Arguments

VAFdata data frame, mutation VAF VAFcov data frame, mutation coverage

minCov numeric, minimum coverage (default: 50)

## Value

data frame, mutation VAF with coverage > minCov

#### References

TBA

filterCommonSNPs

Filter mutations base on coefficient of variant

## **Description**

This function filters common SNPs base on cutoff on coefficient of variant (COV)

## Usage

```
filterCommonSNPs(VAFdata, percentage, output_path)
```

## **Arguments**

VAFdata data frame, mutation VAF

percentage numeric, COV cutoff in percentage (default: 5)
output\_path character, tmp directory to store log file (SNPcv.txt)

#### Value

integer vector, row index of VAF data to be used

filterMultiSources 3

## References

**TBA** 

filterMultiSources Filter multisource contamination

## **Description**

This function filter multisource contamination

## Usage

filterMultiSources(filename,output\_path,VAFdata,VAFcov,uniq\_both)

## **Arguments**

filename character, contamination file name

output\_path character, output directory

VAFdata data frame, mutation VAF

VAFcov data frame, mutation coverage

uniq\_both integer, flag to control type of overlapping mutation in source and target samples

## Value

write to contPredict.txt at output\_path

## References

**TBA** 

inputData Read in data from a file

## Description

This function reads in file with header

## Usage

inputData(filename)

## Arguments

filename character, file name and its path

4 mixingRatio

## Value

data frame

## References

**TBA** 

is\_scalar\_character

Check for scalar argument

## **Description**

This function checks for user's input is a scalar

## Usage

```
is_scalar_character(x)
```

## **Arguments**

Х

character (1)

## **Details**

user's entry must meet the following criteria: character, length 1, not NA, non-zero length

## Value

boolean TRUE or FALSE

mixingRatio

Calculate contamination level

## Description

This function calculates contamination level, ratio of VAF\_target to VAF\_source

## Usage

```
mixingRatio(VAFdata, VAFcov, sample_pairs, final_rel, VAF_cutoff, VAF_ignore,
ALL_flag, output_path, sameSubject)
```

multipleSource 5

#### **Arguments**

VAFdata data frame, mutation VAF
VAFcov data frame, mutation coverage

sample\_pairs data frame, pairwise samples information

final\_rel data frame, relation information

VAF\_cutoff numeric, minimum VAF (default: 0.002)

VAF\_ignore numeric, ignore variant less than this cutoff (default: 0.2)

ALL\_flag logical, TRUE: calculate mixing ratio for all pairwise samples, FALSE: calculate mixing ratio for all pairwise samples for all pairwise samples for all pairwise samples for all pairwise sam

late mixing ratio for contamination pairwise samples

output\_path character, output directory

sameSubject logical, TRUE: calculate contamination level for same subject pairs (default:

FALSE)

#### Value

matrix containing source sample, target sample, relation, contamination level, flip\_flag

#### References

**TBA** 

| multipleSource Identify true source from multisource contamination |
|--------------------------------------------------------------------|
|--------------------------------------------------------------------|

## Description

This function uses Fisher exact test to filter multiple sources that contaminate a particular target sample

#### Usage

```
multipleSource(sample_pairs, VAFdata, VAFcov, VAF_cutoff, VAF_cutoff1, p. val_cutoff, output_path)
```

#### **Arguments**

sample\_pairs data frame, sample infomation
VAFdata data frame, mutation VAF
VAFcov data frame, mutation coverage

VAF\_cutoff numeric, minimum VAF (default: 0.002)

VAF\_cutoff1 numeric, minimum VAF to be considered a source mutation (default: 0.05) p.val\_cutoff numeric, pval cutoff to be considered a significant contamination source (de-

fault: 0.05)

output\_path character, output directory

6 pairCountPoint

## Value

list, remaining paiwise sample relationship information after Fisher exact test that eliminates insignificant contamination pair

#### References

**TBA** 

## **Description**

This function counts number of SNPs in each sample

## Usage

```
numMutationperSample (VAFdata, VAF_cutoff, n)
```

## **Arguments**

VAFdata data frame, mutation VAF
VAF\_cutoff numeric, minimum VAF
n numeric, number of sample

## Value

list, SNP count per sample

#### References

**TBA** 

pairCountPoint Count number of SNPs in 7 regions of a given pair of sample

## **Description**

This function counts number of SNPs in 7 regions of a given pair of sample

## Usage

```
pairCountPoint(VAFdata,pid1,pid2,slope1,slope2)
```

pairPCommon 7

## **Arguments**

VAFdata data frame, mutation VAF
pid1 character, first sample id
pid2 character, second sample id

slope1 numeric, first slope that is closer to axis-X (default: 0.5) slope2 numeric, second slope that is closer to axis-Y (default: 2)

#### Value

matrix, containing number of SNPs in 7 regions

#### References

**TBA** 

pairPCommon Pairwise Sample Function

#### **Description**

4 different functions for pairwise sample

## Usage

```
pairPCommon(VAFdata, VAF_cutoff, num_round_digit, n)
pairShare(VAFdata, VAF_cutoff, n)
pairList(VAFdata, n, del)
pairCommList(SNPcomm, n)
```

## Arguments

VAFdata data frame, mutation VAF VAF\_cutoff numeric, minimum VAF

 ${\tt num\_round\_digit}$ 

numeric, number of rounding decimal digit (default: 3)

n numeric, number of sample

SNPcomm matrix, output from the calling of pairPCommon(VAFdata,VAF\_cutoff,num\_round\_digit,n)

del character, file delimiter

## **Details**

pairPCommon: calculates pcommon

pairShare: counts number of common SNPs pairList: generates all pairwise sample list

pairCommList: tabulates pcommon for all pairwise samples

8 pairRelation

#### Value

pairPCommon: matrix, nxn pcommon, n: number of sample

pairShare: matrix, nxn number of common snps

pairList: vector, all pairwise samples

pairCommList: list, all pairwise samples and their pcommon values

#### References

**TBA** 

pairRelation

Determine relationship between pairwise sample

#### **Description**

This function identifies relationship of pairwise sample [00: same patient | 10: X contaminate Y | 01 Y contaminate X | 11: both way contamination]

## Usage

```
pairRelation(sample_pairs,center_cutoff,source_cutoff,target_cutoff,
localPcomm_cutoff,region_cutoff,num_round_digit,output_path)
```

## **Arguments**

```
sample_pairs
                  data frame, pairwise samples information
center_cutoff
                  numeric, cutoff to be classified as same subject samples (default: 0.5)
source_cutoff
                  numeric, cutoff to be classififed as source sample (default: 0.4)
                  numeric, cutoff to be classified as target sample (default: 0.1)
target_cutoff
localPcomm_cutoff
                  numeric, cutoff to manage high or low SNPs sharing between a pair of samples
                  numeric, cutoff for case with low SNP sharing (default: 0.75)
region_cutoff
num_round_digit
                  integer, rounding numeric up to this decimal point (default: 3)
                  character, output directory
output_path
```

#### Value

matrix, 2 columns containing pcommon and relation

#### References

**TBA** 

regionCountMutation 9

| of samples (s1,s2) | regionCountMutation | Count number of mutation in 7 regions of VAF scatter plot for a pair of samples (s1,s2) |
|--------------------|---------------------|-----------------------------------------------------------------------------------------|
|--------------------|---------------------|-----------------------------------------------------------------------------------------|

## Description

This function counts number of SNPs in 7 regions of pairwise samples VAF scatter plot

#### Usage

```
regionCountMutation(sample_pairs, VAFdata, SNPcount, SNPshare, VAF_cutoff, VAF_ignore, n)
```

#### **Arguments**

sample\_pairs data frame, pairwise samples information

VAFdata data frame, mutation VAF

SNPcount integer matrix, number of SNPs for each sample

SNPshare integer matrix, number of common SNPs
VAF\_cutoff numeric, minimum VAF (default: 0.002)

VAF\_ignore numeric, ignore variant below this cutoff (default: 0.2)

n integer, number of sample

## Value

matrix, (n mutation x 10) containing number of SNPs in 7 regions, number of SNPs in s1, number of SNPs in s2, common SNPs between s1 and s2

#### References

TBA

run\_sampleContamination

Main function to run sample contamination analysis

## **Description**

This function detects sample contamination base on variant allele frequency (VAF) and variant coverage

#### Usage

```
run_sampleContamination(data_path, output_path, config_file="config.txt",
rmSNPcv_cutoff=0,manualsetPara=FALSE,manual_localPcomm=10,manual_center=50,filterCOV=0)
```

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#### **Arguments**

data\_path character, mutation VAF and coverage file directory

output\_path character, output directory config\_file character, configuration file

rmSNPcv\_cutoff numeric, TRUE: filter SNPs with low covariance of coefficient (COV) (default:

FALSE)

manualsetPara logical, TRUE: manual setting base on distribution of pairwise samples com-

monality (default: FALSE)

manual\_localPcomm

numeric, manual setting of localPcomm for low or high contamination (default:

10)

manual\_center numeric, manual setting of center cutoff for same subject determination (default:

50)

filterCOV numeric, filter mutation below this cutoff

#### Value

list, containing pcomm, center cutoff, target cutoff, source cutoff, region cutoff and number of sample

#### References

TBA

setParameterConfig Set parameters base on configuration file

#### **Description**

This function takes in data from configuration file to set parameters

#### Usage

setParameterConfig(configFile)

#### **Arguments**

configFile character, configuration file name and its path

#### References

TBA

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