Package 'TPES'

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Title Tumor Purity Estimation using SNVs
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Description A bioinformatics tool for the estimation of the tumor purity from sequencing data. It uses the set of putative clonal somatic single nucleotide variants within copy number neutral segments to call tumor cellularity.
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TCGA_A8_A0A7_maf

SNVsReadCountsFile for sample TCGA-A8-A0A7

Description

A data frame object containing the read counts data of somatic single nucleotide variants (SNVs) loci for sample TCGA-A8-A0A7. The header contains the chromosme that harbors the SNV ("chr" column), the position of the SNV (defined by the "start" and "end" columns), the informations about the reference and alternative base counts ("ref.count" and "alt.count" columns, respectively) and finally the sample ID ("sample" column). For more information please visit MAF file format.

Format

A data.frame object.

TCGA_A8_A0A7_ploidy

Ploidy data for sample TCGA-A8-A0A7

Description

A data frame containing the ploidy status of a sample. It must contains at least the sample ID ("sample" column) and the ploidy status ("ploidy" column).

Format

A data.frame object.

TCGA_A8_A0A7_seg

SEG file (segmented data) for sample TCGA-A8-A0A7

Description

A data frame object that lists loci and associated numeric values. The header must be compatible with the standard format defined by the Broad Institute. For more information please visit SEG file format.

Format

A data.frame object.

TCGA_HT_8564_maf

SNVsReadCountsFile for sample TCGA-HT-8564

Description

A data frame object containing the read counts data of somatic single nucleotide variants (SNVs) loci for sample TCGA-HT-8564. The header contains the chromosme that harbors the SNV ("chr" column), the position of the SNV (defined by the "start" and "end" columns), the informations about the reference and alternative base counts ("ref.count" and "alt.count" columns, respectively) and finally the sample ID ("sample" column). For more information please visit MAF file format.

Format

A data.frame object.

TCGA_HT_8564_ploidy

Ploidy data for sample TCGA-HT-8564

Description

A data frame containing the ploidy status of a sample. It must contains at least the sample ID ("sample" column) and the ploidy status ("ploidy" column).

Format

A data.frame object.

TCGA_HT_8564_seg

SEG file (segmented data) for sample TCGA-HT-8564

Description

A data frame object that lists loci and associated numeric values. The header must be compatible with the standard format defined by the Broad Institute. For more information please visit SEG file format.

Format

A data.frame object.

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Tumor Purity Estimation using SNVs

Description

TPES_purity function estimates tumor purity.

Usage

```
TPES_purity(ID, SEGfile, SNVsReadCountsFile, ploidy, RMB = 0.47,
 maxAF = 0.55, minCov = 10, minAltReads = 5, minSNVs = 10)
```

Arguments

ΙD Sample ID. Must be the same ID as in SEGfile, SNVsReadCountsFile and

ploidy.

SEGfile A standard SEG file (segmented data). It is a data frame object that lists loci and

> associated numeric values. The header must be compatible with the standard format defined by the Broad Institute. For more information please visit SEG

file format.

SNVsReadCountsFile

A standard MAF (Mutation Annotation Format) file. It is a data frame object containing the read counts data of somatic single nucleotide variants (SNVs) loci. The header must contains at least informations about the chromosme that harbors the SNV ("chr" column), the position of the SNV (defined by the "start" and "end" columns), the sample ID ("sample" column) and finally the informations about the reference and alternative base counts ("ref.count" and "alt.count" columns, respectively). For more information please visit MAF file format.

ploidy A data frame containing the ploidy status of a sample. It must contain at least

the sample ID ("sample" column) and the ploidy status ("ploidy" column).

RMB The Reference Mapping Bias value. The reference genome contains only one

> allele at any given locus, so reads that carry a non-reference allele are less likely to be mapped during alignment; this causes a shift from 0.5. It can be estimated as: 1 - medAF, where medAF is the median value of the allelic fraction of the sample's germline heterozygous SNPs. Default is set to 0.47. For more

informations see: PMID: 19808877.

The filter on the allelic fraction (AF) distribution of SNVs. This is necessary to maxAF

be sure to keep only heterozygous SNVs. Clonal and subclonal SNVs, which

have an AF greater than maxAF, will be removed.

The minimum coverage for a SNV to be retained.

minAltReads The minimum coverage for the alternative base of a SNV to be retained.

minSNVs The minimum number of SNVs required to make a purity call.

minCov

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Value

TPES returns a data.frame object with one row per sample and the following columns:

sample	The sample ID;
purity	The sample purity estimated by TPES;
purity.min	The sample minimum purity estimated by TPES;
purity.max	The sample maximum purity estimated by TPES;
n.segs	The number of copy number neutral segments used by TPES;
n.SNVs	The number of SNVs used by TPES;
RMB	The Reference Mapping Bias value used to estimate the tumor purity;
BandWidth	The smoothing bandwidth value of the density function chosen by TPES.
log	Reports if the run was successful; otherwise provides debugging information.

Examples

```
## Compute tumor purity for samples "TCGA-A8-A0A7" and "TCGA-HT-8564"
## https://cancergenome.nih.gov/
## Please copy and paste the following lines:
library(TPES)
TPES_purity(ID = "TCGA-A8-A0A7", SEGfile = TCGA_A8_A0A7_seg,
SNVsReadCountsFile = TCGA_A8_A0A7_maf, ploidy = TCGA_A8_A0A7_ploidy,
RMB = 0.47, maxAF = 0.55, minCov = 10, minAltReads = 5, minSNVs = 10)
TPES_purity(ID = "TCGA-HT-8564", SEGfile = TCGA_HT_8564_seg,
SNVsReadCountsFile = TCGA_HT_8564_maf, ploidy = TCGA_HT_8564_ploidy,
RMB = 0.47, maxAF = 0.55, minCov = 10, minAltReads = 5, minSNVs = 10)
```

TPES_report

Tumor Purity Estimation using SNVs

Description

TPES_report function produces a graphical report regarding the allelic fraction values of the putative clonal SNVs used by TPES_purity and the density function(s) computed by TPES_purity.

Usage

```
TPES_report(ID, SEGfile, SNVsReadCountsFile, ploidy, RMB = 0.47,
    maxAF = 0.55, minCov = 10, minAltReads = 5, minSNVs = 10)
```

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Arguments

ID Sample ID. Must be the same ID as in SEGfile, SNVsReadCountsFile and

ploidy.

SEGfile A standard SEG file (segmented data). It is a data frame object that lists loci and

associated numeric values. The header must be compatible with the standard format defined by the Broad Institute. For more information please visit SEG

file format.

SNVsReadCountsFile

A standard MAF (Mutation Annotation Format) file. It is a data frame object containing the read counts data of somatic single nucleotide variants (SNVs) loci. The header must contains at least informations about the chromosme that harbors the SNV ("chr" column), the position of the SNV (defined by the "start" and "end" columns), the sample ID ("sample" column) and finally the informations about the reference and alternative base counts ("ref.count" and "alt.count" columns, respectively). For more information please visit MAF file format.

ploidy A data frame containing the ploidy status of a sample. It must contain at least

the sample ID ("sample" column) and the ploidy status ("ploidy" column).

RMB The Reference Mapping Bias value. The reference genome contains only one

allele at any given locus, so reads that carry a non-reference allele are less likely to be mapped during alignment; this causes a shift from 0.5. It can be estimated as: 1-medAF, where medAF is the median value of the allelic fraction of the sample's germline heterozygous SNPs. Default is set to 0.47. For more

informations see: PMID: 19808877.

maxAF The filter on the allelic fraction (AF) distribution of SNVs. This is necessary to

be sure to keep only heterozygous SNVs. Clonal and subclonal SNVs, which

have an AF greater than maxAF, will be removed.

minCov The minimum coverage for a SNV to be retained.

minAltReads The minimum coverage for the alternative base of a SNV to be retained.

minSNVs The minimum number of SNVs required to make a purity call.

Value

A plot with:

histogram Represents the allelic fraction distribution of putative clonal and subclonal (if

presents) SNVs within copy number neutral segments and the peak(s) detected

by TPES;

density plot Represents how the density function varies according to different bandwidth

values (for more information see density); only the bandwidth values that result

in at most 2 peaks are considered.

Examples

```
## Generate TPES report for samples "TCGA-A8-A0A7" and "TCGA-HT-8564"
## https://cancergenome.nih.gov/
## Please copy and paste the following lines:
library(TPES)
```

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```
TPES_report(ID = "TCGA-A8-A0A7", SEGfile = TCGA_A8_A0A7_seg,
SNVsReadCountsFile = TCGA_A8_A0A7_maf, ploidy = TCGA_A8_A0A7_ploidy,
RMB = 0.47, maxAF = 0.55, minCov = 10, minAltReads = 5, minSNVs = 10)
```

TPES_report(ID = "TCGA-HT-8564", SEGfile = TCGA_HT_8564_seg,
SNVsReadCountsFile = TCGA_HT_8564_maf, ploidy = TCGA_HT_8564_ploidy,
RMB = 0.47, maxAF = 0.55, minCov = 10, minAltReads = 5, minSNVs = 10)

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