Package 'ASCAT'

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Description

run ASPCF segmentation

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Usage

```
ascat.aspcf(ASCATobj, selectsamples = 1:length(ASCATobj$samples),
ascat.gg = NULL, penalty = 25)
```

Arguments

ASCATobj an ASCAT object selectsamples

a vector containing the sample number(s) to PCF. Default = all

ascat.gg germline genotypes (NULL if germline data is available)

penalty penalty of introducing an additional ASPCF breakpoint (expert parameter, don't

adapt unless you know what you're doing)

Details

This function can be easily parallelised by controlling the selectsamples parameter it saves the results in LogR_PCFed[sample]_[segment].txt and BAF_PCFed[sample]_[segment].txt; if these files exist, the results are read from them.

Hence, after parallelisation, copy all the files into the current directory, and run this function to read in the results

Value

output: ascat data structure containing:

- 1. Tumor_LogR data matrix
- 2. Tumor_BAF data matrix
- 3. Tumor_LogR_segmented: matrix of LogR segmented values
- 4. Tumor_BAF_segmented: list of BAF segmented values; each element in the list is a matrix containing the segmented values for one sample (only for probes that are germline homozygous)
- 5. Germline_LogR data matrix
- 6. Germline BAF data matrix
- 7. SNPpos: position of all SNPs
- 8. ch: a list containing vectors with the indices for each chromosome (e.g. Tumor_LogR[ch[[13]],] will output the Tumor_LogR data of chromosome 13
- 9. chr: a list containing vectors with the indices for each distinct part that can be segmented separately (e.g. chromosome arm, stretch of DNA between gaps in the array design)

ascat.GCcorrect ascat.GCcorrect

Description

Corrects logR of the tumour sample(s) with genomic GC content

Usage

```
ascat.GCcorrect(ASCATobj, GCcontentfile = NULL)
```

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Arguments

```
ASCATobj an ASCAT object GCcontentfile
```

File containing the GC content around every SNP for increasing window sizes

Details

Note that probes not present in the GCcontentfile will be lost from the results

Value

ASCAT object with corrected tumour logR

```
ascat.loadData a
```

ascat.loadData

Description

Function to read in SNP array data

Usage

```
ascat.loadData(Tumor_LogR_file, Tumor_BAF_file, Germline_LogR_file = NULL,
   Germline_BAF_file = NULL, chrs = c(1:22, "X", "Y"), gender = NULL,
   sexchromosomes = c("X", "Y"))
```

Arguments

Details

germline data files can be NULL - in that case these are not read in

Value

ascat data structure containing:

- 1. Tumor_LogR data matrix
- 2. Tumor_BAF data matrix
- 3. Tumor_LogR_segmented: placeholder, NULL
- 4. Tumor_BAF_segmented: placeholder, NULL
- 5. Germline_LogR data matrix
- 6. Germline_BAF data matrix
- 7. SNPpos: position of all SNPs
- 8. ch: a list containing vectors with the indices for each chromosome (e.g. Tumor_LogR[ch[[13]],] will output the Tumor_LogR data of chromosome 13
- 9. chr: a list containing vectors with the indices for each distinct part that can be segmented separately (e.g. chromosome arm, stretch of DNA between gaps in the array design)
- 10. gender: a vector of gender for each cases ("XX" or "XY"). Default = NULL: all female ("XX")

```
ascat.plotAscatProfile
```

ascat.plotAscatProfile

Description

Function plotting the rounded ASCAT profiles over all chromosomes

Usage

```
ascat.plotAscatProfile(n1all, n2all, heteroprobes, ploidy, rho, goodnessOfFit,
nonaberrant, y_limit = 5, ch, lrr, bafsegmented)
```

Arguments

n1all copy number major allele n2all copy number minor allele

heteroprobes probes with heterozygous germline

ploidy ploidy of the sample rho purity of the sample

goodnessOfFit

estimated goodness of fit

nonaberrant boolean flag denoting non-aberrated samples

y_limit Optional parameter determining the size of the y axis in the nonrounded plot and

ASCAT profile. Default=5

ch a list containing c vectors, where c is the number of chromosomes and every

vector contains all probe numbers per chromosome

1rr (unsegmented) log R, in genomic sequence (all probes), with probe IDs

 $\verb|bafsegmented| B Allele Frequency, segmented, in genomic sequence (only probes heterozygous$

in germline), with probe IDs

Value

plot showing the ASCAT profile of the sample

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```
ascat.plotGenotypes
```

ascat.plotGenotypes

Description

ascat.plotGenotypes

Usage

```
ascat.plotGenotypes(ASCATobj, title, Tumor_BAF_noNA, Hom, ch_noNA)
```

Arguments

```
ASCATobj an ASCAT object title main title of the plot
```

Tumor_BAF_noNA

B-allele frequencies of the tumour sample with removed NA values

Hom Boolean vector denoting homozygous SNPs

ch_noNA vector of probes per chromosome (NA values excluded)

Value

plot showing classified BAF per sample, with unused SNPs in green, germline homozygous SNPs in blue and all others in red

```
ascat.plotNonRounded
```

ascat.plotNonRounded

Description

Function plotting the unrounded ASCAT copy number over all chromosomes

Usage

```
ascat.plotNonRounded(ploidy, rho, goodnessOfFit, nonaberrant, nAfull, nBfull,
    y_limit = 5, bafsegmented, ch, lrr)
```

Arguments

```
ploidy ploidy of the sample rho purity of the sample
```

goodnessOfFit

estimated goodness of fit

nonaberrant boolean flag denoting non-aberrated samples

nAfull copy number major allele

nBfull copy number minor allele

y_limit Optional parameter determining the size of the y axis in the nonrounded plot and

ASCAT profile. Default=5

bafsegmented B Allele Frequency, segmented, in genomic sequence (only probes heterozygous

in germline), with probe IDs

ch a list containing c vectors, where c is the number of chromosomes and every

vector contains all probe numbers per chromosome

1rr (unsegmented) log R, in genomic sequence (all probes), with probe IDs

Value

plot showing the nonrounded copy number profile, using base plotting function

Description

Plots SNP array data

Usage

```
ascat.plotRawData(ASCATobj)
```

Arguments

ASCATobj an ASCAT object (e.g. data structure from ascat.loadData)

Value

Produces png files showing the logR and BAF values for tumour and germline samples

Description

plots the SNP array data before and after segmentation

Usage

```
ascat.plotSegmentedData(ASCATobj)
```

Arguments

```
ASCATobj an ASCAT object (e.g. from ascat.aspcf)
```

Value

png files showing raw and segmented tumour logR and BAF

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```
\verb"ascat.plotSunrise" \textit{ascat.plotSunrise}
```

Description

ascat.plotSunrise

Usage

```
ascat.plotSunrise(d, psi_opt1, rho_opt1, minim = T)
```

Arguments

d distance matrix for a range of ploidy and tumour percentage values

rho_opt1 optimal aberrant cell fraction

minim when set to true, optimal regions in the sunrise plot are depicted in blue; if set

to false, colours are inverted and red corresponds to optimal values (default:

TRUE)

Value

plot visualising range of ploidy and tumour percentage values

```
as cat. \verb|predictGermlineGenotypes| \\ as cat. \verb|predictGermlineGenotypes|
```

Description

predicts the germline genotypes of samples for which no matched germline sample is available

Usage

```
ascat.predictGermlineGenotypes(ASCATobj, platform = "AffySNP6")
```

Arguments

```
ASCATobj an ASCAT object platform used array platform
```

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Details

Currently possible values for platform:

AffySNP6 (default)

Custom10k

Illumina109k

Illumina Cyto SNP

Illumina610k

Illumina660k

Illumina700k

 $Illumina \\ 1 \\ M$

Illumina2.5M IlluminaOmni5

Affy10k

Affy100k

Affy250k_sty

Affy250k_nsp

AffyOncoScan

AffyCytoScanHD

HumanCNV370quad

HumanCore12

Human Core Exome 24

HumanOmniExpress12

Value

predicted germline genotypes

ascat.runAscat

ascat.runAscat

Description

ASCAT main function, calculating the allele-specific copy numbers

Usage

```
ascat.runAscat(ASCATobj, gamma = 0.55, pdfPlot = F, y_limit = 5,
 circos = NA, rho_manual = NA, psi_manual = NA)
```

Arguments

ASCATobj	an ASCAT object from ascat.aspcf
gamma	technology parameter, compaction of Log R profiles (expected decrease in case of deletion in diploid sample, 100% aberrant cells; 1 in ideal case, 0.55 of Illumina $109K$ arrays)
pdfPlot	Optional flag if nonrounded plots and ASCAT profile in pdf format are desired. Default=F
y_limit	Optional parameter determining the size of the y axis in the nonrounded plot and ASCAT profile. Default=5

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circos	Optional file to output the non-rounded values in Circos track format. Default=NA
rho_manual	optional argument to override ASCAT optimization and supply rho parameter (not recommended)
psi_manual	optional argument to override ASCAT optimization and supply psi parameter (not recommended)

Details

Note: for copy number only probes, nA contains the copy number value and nB = 0.

Value

an ASCAT output object, containing:

- 1. nA: copy number of the A allele
- 2. nB: copy number of the B allele
- 3. aberrantcellfraction: the aberrant cell fraction of all arrays
- 4. ploidy: the ploidy of all arrays
- 5. failedarrays: arrays on which ASCAT analysis failed
- 6. nonaberrantarrays: arrays on which ASCAT analysis indicates that they show virtually no aberrations
- 7. segments: an array containing the copy number segments of each sample (not including failed arrays)
- 8. segments_raw: an array containing the copy number segments of each sample without any rounding applied