Package 'ASCAT'

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Type Package

Title Allele-Specific Copy Number Analysis of Tumours

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Description R package of ASCAT as published in http://www.ncbi.nlm.nih.gov/pubmed/20837533

Depends R (>= 2.13.0),
RColorBrewer,
grDevices,
graphics,
stats,
utils

License GPL-3

LazyLoad yes

RoxygenNote 7.1.2

R topics documented:

`ascat.asmultipcf` *Allele-specific segmentation of multiple samples*

Description

This segmentation function should only be used if part of the breakpoints are expected to be shared between samples, e.g. due to a common ancestry.

Usage

```
ascat.asmultipcf(
  ASCATobj,
  ascat.gg = NULL,
  penalty = 25,
  wsample = NULL,
  selectAlg = "exact",
  refine = TRUE
)
```

Arguments

`ASCATobj`	an ASCAT object
`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 are doing)
`wsample`	Vector of length length(ASCATobj$samples). Can be used to assign different weights to samples, for example to account for differences in sequencing quality. (Default = NULL)
`selectAlg`	Set to "exact" to run the exact algorithm, or "fast" to run the heuristic algorithm. (Default = "exact")
`refine`	Logical. Should breakpoints be refined on a per sample base? Otherwise each breakpoint is assumed to be present in each sample. (Default = TRUE)

Details

This function saves the results in in [sample].LogR.PCFed.txt and [sample].BAF.PCFed.txt

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.aspcf` *ascat.aspcf*

Description

run ASPCF segmentation

Usage

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

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)
`out.dir`	directory in which output files will be written
`out.prefix`	prefix for output file names

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

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 not 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)
```

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` *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

`Tumor_LogR_file`	file containing logR of tumour sample(s)
`Tumor_BAF_file`	file containing BAF of tumour sample(s)
`Germline_LogR_file`	file containing logR of germline sample(s), NULL
`Germline_BAF_file`	file containing BAF of germline sample(s), NULL
`chrs`	a vector containing the names for the chromosomes (e.g. c(1:22,"X"))
`gender`	a vector of gender for each cases ("XX" or "XY"). Default = all female ("XX")
`sexchromosomes`	a vector containing the names for the sex chromosomes

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,
  chrs
)
```

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
lrr	(unsegmented) log R, in genomic sequence (all probes), with probe IDs
bafsegmented	B Allele Frequency, segmented, in genomic sequence (only probes heterozygous in germline), with probe IDs
chrs	a vector containing the names for the chromosomes (e.g. c(1:22,"X"))

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

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`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,
  chrs
)
```

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
`lrr`	(unsegmented) log R, in genomic sequence (all probes), with probe IDs
`chrs`	a vector containing the names for the chromosomes (e.g. c(1:22,"X"))

Value

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

ascat.plotRawData *ascat.plotRawData*

Description

Plots SNP array data

Usage

```
ascat.plotRawData(ASCATobj, img.dir = ".", img.prefix = "")
```

Arguments

ASCATobj	an ASCAT object (e.g. data structure from ascat.loadData)
img.dir	directory in which figures will be written
img.prefix	prefix for figure names

Value

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

ascat.plotSegmentedData *ascat.plotSegmentedData*

Description

plots the SNP array data before and after segmentation

Usage

```
ascat.plotSegmentedData(ASCATobj, img.dir = ".", img.prefix = "")
```

Arguments

ASCATobj	an ASCAT object (e.g. from ascat.aspcf)
img.dir	directory in which figures will be written
img.prefix	prefix for figure names

Value

png files showing raw and segmented tumour logR and BAF

`ascat.plotSunrise` *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
`psi_opt1`	optimal ploidy
`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

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`ascat.predictGermlineGenotypes` *ascat.predictGermlineGenotypes*

Description

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

Usage

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

Arguments

`ASCATobj`	an ASCAT object
`platform`	used array platform
`img.dir`	directory in which figures will be written
`img.prefix`	prefix for figure names

Details

Currently possible values for platform:
AffySNP6 (default)
Custom10k
IlluminaASA
IlluminaGSAv3
Illumina109k
IlluminaCytoSNP
IlluminaCytoSNP850k
Illumina610k
Illumina660k
Illumina700k
Illumina1M
Illumina2.5M
IlluminaOmni5
Affy10k
Affy100k
Affy250k_sty
Affy250k_nsp
AffyOncoScan
AffyCytoScanHD
HumanCNV370quad
HumanCore12
HumanCoreExome24
HumanOmniExpress12
IlluminaOmniExpressExome

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,
  img.dir = ".",
  img.prefix = ""
)
```

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
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)
img.dir	directory in which figures will be written
img.prefix	prefix for figure names

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
9. distance_matrix: distances for a range of ploidy and tumor percentage values

Index