Package ‘cn.mops’

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License LGPL (>= 2.0)
Type Package
Title cn.mops - Mixture of Poissons for CNV detection in NGS data
Description cn.mops (Copy Number estimation by a Mixture Of PoissonS) is a data processing pipeline for copy number variations and aberrations (CNVs and CNAs) from next generation sequencing (NGS) data. The package supplies functions to convert BAM files into read count matrices or genomic ranges objects, which are the input objects for cn.mops. cn.mops models the depths of coverage across samples at each genomic position. Therefore, it does not suffer from read count biases along chromosomes. Using a Bayesian approach, cn.mops decomposes read variations across samples into integer copy numbers and noise by its mixture components and Poisson distributions, respectively. cn.mops guarantees a low FDR because wrong detections are indicated by high noise and filtered out. cn.mops is very fast and written in C++.
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calcFractionalCopyNumbers

Description

This generic function calculates the fractional copy numbers of a CNV detection method stored in an instance of \texttt{CNVDetectionResult-class}. Must be a result of "referencecn.mops".

Arguments

- \texttt{object}: An instance of "CNVDetectionResult"
- \texttt{segStat}: Which statistic per segment should be used. Can be either "mean" or "median". (Default="mean").

Value

\texttt{calcFractionalCopyNumbers} returns an instance of "CNVDetectionResult".

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

\begin{verbatim}
data(cn.mops)
r <- referencecn.mops(X[,1:2],apply(X,1,median))
calcFractionalCopyNumbers(r)
\end{verbatim}

Usage

\begin{verbatim}
## S4 method for signature 'CNVDetectionResult'
calcFractionalCopyNumbers(object,
  segStat = "mean")
\end{verbatim}

Arguments

- \texttt{object}: An instance of "CNVDetectionResult"
- \texttt{segStat}: Which statistic per segment should be used. Can be either "mean" or "median". (Default="mean").
calcIntegerCopyNumbers

Value

calcFractionalCopyNumbers returns an instance of "CNVDetectionResult".

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- referencecn.mops(X[,1:2],apply(X,1,median))
calcFractionalCopyNumbers(r)

calcIntegerCopyNumbers

Calculation of integer copy numbers for the CNVs and CNV regions.

Description

This generic function calculates the integer copy numbers of a CNV detection method stored in an instance of CNVDetectionResult-class.

Arguments

object An instance of "CNVDetectionResult"

Value

calcIntegerCopyNumbers returns an instance of "CNVDetectionResult".

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
calcIntegerCopyNumbers(r)
Description

This generic function calculates the integer copy numbers of a CNV detection method stored in an instance of \texttt{CNVDetectionResult-class}.

Usage

```r
## S4 method for signature 'CNVDetectionResult'
calcIntegerCopyNumbers(object)
```

Arguments

- \texttt{object} An instance of "CNVDetectionResult"

Value

\texttt{calcIntegerCopyNumbers} returns an instance of "CNVDetectionResult".

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
calcIntegerCopyNumbers(r)
```

\texttt{cn.mops} \hspace{1cm} \textit{Copy number detection in NGS data.}

Description

This function performs the \texttt{cn.mops} algorithm for copy number detection in NGS data.

Usage

```r
cn.mops(input, I = c(0.025, 0.5, 1, 1.5, 2, 2.5, 3, 3.5, 4),
classes = c("CN0", "CN1", "CN2", "CN3", "CN4", "CN5", "CN6", "CN7", "CN8"),
priorImpact = 1, cyc = 20, parallel = 0, norm = 1,
normType = "poisson", sizeFactor = "mean", normQu = 0.25,
quSizeFactor = 0.75, upperThreshold = 0.5, lowerThreshold = -0.9,
minWidth = 3, segAlgorithm = "fast", minReadCount = 5,
useMedian = FALSE, returnPosterior = FALSE, ...)
```
Arguments

input
Either an instance of "GRanges" or a raw data matrix, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region.

I
Vector positive real values that contain the expected fold change of the copy number classes. Length of this vector must be equal to the length of the "classes" parameter vector. For human copy number polymorphisms we suggest to use the default \( I = c(0.025,0.5,1,1.5,2,2.5,3,3.5,4) \).

classes
Vector of characters of the same length as the parameter vector "I". One vector element must be named "CN2". The names reflect the labels of the copy number classes. Default = c("CN0","CN1","CN2","CN3","CN4","CN5","CN6","CN7","CN8").

priorImpact
Positive real value that reflects how strong the prior assumption affects the result. The higher the value the more samples will be assumed to have copy number 2. Default = 1.

cyc
Positive integer that sets the number of cycles for the algorithm. Usually after less than 15 cycles convergence is reached. Default = 20.

parallel
How many cores are used for the computation. If set to zero than no parallelization is applied. Default = 0.

norm
The normalization strategy to be used. If set to 0 the read counts are not normalized and cn.mops does not model different coverages. If set to 1 the read counts are normalized. If set to 2 the read counts are not normalized and cn.mops models different coverages. (Default=1).

normType
Mode of the normalization technique. Possible values are "mean","min","median","quant","poisson" and "mode". Read counts will be scaled sample-wise. Default = "poisson".

sizeFactor
By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").

normQu
Real value between 0 and 1. If the "normType" parameter is set to "quant" then this parameter sets the quantile that is used for the normalization. Default = 0.25.

quSizeFactor
Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization". Real value between 0 and 1. Default = 0.75.

upperThreshold
Positive real value that sets the cut-off for copy number gains. All CNV calling values above this value will be called as "gain". The value should be set close to the log2 of the expected foldchange for copy number 3 or 4. Default = 0.5.

lowerThreshold
Negative real value that sets the cut-off for copy number losses. All CNV calling values below this value will be called as "loss". The value should be set close to the log2 of the expected foldchange for copy number 1 or 0. Default = -0.9.

minWidth
Positive integer that is exactly the parameter "min.width" of the "segment" function of "DNAcopy". minWidth is the minimum number of segments a CNV should span. Default = 3.

segAlgorithm
Which segmentation algorithm should be used. If set to "DNAcopy" circular binary segmentation is performed. Any other value will initiate the use of our fast segmentation algorithm. Default = "fast".

minReadCount
If all samples are below this value the algorithm will return the prior knowledge. This prevents that the algorithm from being applied to segments with very low coverage. Default=5.
useMedian     Whether "median" instead of "mean" of a segment should be used for the CNV call. Default=FALSE.
returnPosterior     Flag that decides whether the posterior probabilities should be returned. The posterior probabilities have a dimension of samples times copy number states times genomic regions and therefore consume a lot of memory. Default=FALSE.

Additional parameters will be passed to the "DNAcopy" or the standard segmentation algorithm.

Value
An instance of "CNVDetectionResult".

Author(s)
Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
cn.mops(XRanges)
cn.mops(XRanges,parallel=2)
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Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

showClass("CNVDetectionResult")

cnvr

This generic function returns CNV regions of a CNV detection method stored in an instance of `CNVDetectionResult`-class.

Description

This generic function returns CNV regions of a CNV detection method stored in an instance of `CNVDetectionResult`-class.

Arguments

<table>
<thead>
<tr>
<th>Argument</th>
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<tbody>
<tr>
<td>object</td>
<td>An instance of &quot;CNVDetectionResult&quot;</td>
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</tbody>
</table>

Value

cnvr returns a "GRanges" object containing the CNV regions.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>
Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
cnvr(r)

---

cnvr,CNVDetectionResult-method

This generic function returns CNV regions of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Description

This generic function returns CNV regions of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Usage

## S4 method for signature 'CNVDetectionResult'
cnvr(object)

Arguments

object An instance of "CNVDetectionResult"

Value

cnvr returns a "GRanges" object containing the CNV regions.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
cnvr(r)

---

CNVRanges Genomic locations and indices of the simulated CNVs.

Description

This data set gives the starts, ends, and the integer copy number of the simulated CNVs in the data set `XRanges` object.

Usage

CNVRanges
cnvs

Format

A GRanges object with 20 rows and 40 value columns across 1 space.

Source

http://www.bioinf.jku.at/cnmops/cnmops.html.

References


cnvs

This generic function returns CNVs of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Description

This generic function returns CNVs of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Arguments

object

An instance of "CNVDetectionResult"

Value

cnvs returns a GRanges object containing the CNVs.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
cnvs(r)
Description

This generic function returns CNVs of a CNV detection method stored in an instance of \texttt{CNVDetectionResult-class}.

Usage

\texttt{## S4 method for signature 'CNVDetectionResult'
\texttt{cnvs(object)}

Arguments

\texttt{object} An instance of "CNVDetectionResult"

Value

cnvs returns a GRanges object containing the CNVs.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
cnvs(r)

Description

Copy number detection in exome sequencing data.

Usage

\texttt{exomecn.mops(input, I = c(0.025, 0.5, 1, 1.5, 2, 2.5, 3, 3.5, 4),
classes = c("CN0", "CN1", "CN2", "CN3", "CN4", "CN5", "CN6", "CN7", "CN8"),
priorImpact = 10, cyc = 20, parallel = 0, norm = 1,
normType = "poisson", sizeFactor = "mean", normQu = 0.25,
quSizeFactor = 0.75, upperThreshold = 0.5, lowerThreshold = -0.8,
minWidth = 5, segAlgorithm = "fast", minReadCount = 1,
useMedian = FALSE, returnPosterior = FALSE, ...)}
Arguments

input Either an instance of "GRanges" or a raw data matrix, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region.

I Vector positive real values that contain the expected fold change of the copy number classes. Length of this vector must be equal to the length of the "classes" parameter vector. For human copy number polymorphisms we suggest to use the default $I = c(0.025,0.5,1,1.5,2,2.5,3,3.5,4)$.

classes Vector of characters of the same length as the parameter vector "I". One vector element must be named "CN2". The names reflect the labels of the copy number classes. Default = c("CN0","CN1","CN2","CN3","CN4","CN5","CN6","CN7","CN8").

priorImpact Positive real value that reflects how strong the prior assumption affects the result. The higher the value the more samples will be assumed to have copy number 2. Default = 10.

cyc Positive integer that sets the number of cycles for the algorithm. Usually after less than 15 cycles convergence is reached. Default = 20.

parallel How many cores are used for the computation. If set to zero than no parallelization is applied. Default = 0.

norm The normalization strategy to be used. If set to 0 the read counts are not normalized and cn.mops does not model different coverages. If set to 1 the read counts are normalized. If set to 2 the read counts are not normalized and cn.mops models different coverages. (Default=1).

normType Mode of the normalization technique. Possible values are "mean","min","median","quant", "poisson" and "mode". Read counts will be scaled sample-wise. Default = "poisson".

sizeFactor By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").

normQu Real value between 0 and 1. If the "normType" parameter is set to "quant" then this parameter sets the quantile that is used for the normalization. Default = 0.25.

quSizeFactor Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization". Real value between 0 and 1. Default = 0.75.

upperThreshold Positive real value that sets the cut-off for copy number gains. All CNV calling values above this value will be called as "gain". The value should be set close to the log2 of the expected foldchange for copy number 3 or 4. Default = 0.55.

lowerThreshold Negative real value that sets the cut-off for copy number losses. All CNV calling values below this value will be called as "loss". The value should be set close to the log2 of the expected foldchange for copy number 1 or 0. Default = -0.8.

minWidth Positive integer that is exactly the parameter "min.width" of the "segment" function of "DNAcopy". minWidth is the minimum number of segments a CNV should span. Default = 5.

segAlgorithm Which segmentation algorithm should be used. If set to "DNAcopy" circular binary segmentation is performed. Any other value will initiate the use of our fast segmentation algorithm. Default = "fast".

minReadCount If all samples are below this value the algorithm will return the prior knowledge. This prevents that the algorithm from being applied to segments with very low coverage. Default=1.
exomeCounts

useMedian Whether "median" instead of "mean" of a segment should be used for the CNV call. Default=FALSE.

returnPosterior Flag that decides whether the posterior probabilities should be returned. The posterior probabilities have a dimension of samples times copy number states times genomic regions and therefore consume a lot of memory. Default=FALSE.

... Additional parameters will be passed to the "DNAcopy" or the standard segmentation algorithm.

Value An instance of "CNVDetectionResult".

Author(s) Guenter Klambauer <klambauer@bioinf.jku.at>

Examples
data(cn.mops)
exomecn.mops(exomeCounts)

Description This data set gives the read counts on chromosome 22 (hg19) of 22 samples in 3785 exons. The rows correspond to targeted regions or exons and columns to samples. An entry is the number of reads that map to the specific segment, i.e. targeted region or exon, of the sample. The GRanges object contains the information of the genomic location. The read counts were generated from freely available exome sequencing data of the 1000Genomes Project.

Usage exomeCounts

Format A GRanges object of 3785 rows and 22 columns.

getReadCountsFromBAM

Description

Calculates read counts from BAM files. These counts are necessary for CNV detection methods based on depth of coverage information. Note that the function is much faster, if the BAM files have an index file. The index file is assumed to be in the same folder and have an identical file name except that ".bai" is appended.

This function can also be run in a parallel version.

Usage

getReadCountsFromBAM(BAMFiles, sampleNames, refSeqName, WL, mode, parallel = 0)

Arguments

- **BAMFiles**: BAMFiles
- **sampleNames**: The corresponding sample names to the BAM Files.
- **refSeqName**: Name of the reference sequence that should be analyzed. The name must appear in the header of the BAM file. If it is not given, cn.mops will select the first reference sequence that appears in the header of the BAM files.
- **WL**: Windowlength. Length of the initial segmentation of the genome in basepairs. Should be chosen such that on the average 100 reads are contained in each segment. If not given, cn.mops will try to find an appropriate window length.
- **mode**: Possible values are "paired" and "unpaired", whether the mapping algorithm was using a "paired" or "unpaired" strategy.
- **parallel**: The number of parallel processes to be used for this function. Default=0.

Value

An instance of "GRanges", that contains the breakpoints of the initial segments and the raw read counts that were extracted from the BAM files. This object can be used as input for cn.mops and other CNV detection methods.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>
getSegmentReadCountsFromBAM

Calculation of read counts from BAM files for predefined segments.

Description
Generates the read counts from BAM Files for predefined segments. This is the appropriate choice for exome sequencing data, where the bait regions, target regions or exons are the predefined segments. These counts are necessary for CNV detection methods based on depth of coverage information. Note that the function is much faster, if the BAM files have an index file. The index file is assumed to be in the same folder and have an identical file name except that ".bai" is appended. This function can also be run in a parallel version.

Usage
getSegmentReadCountsFromBAM(BAMFiles, GR, sampleNames, mode, parallel = 0, BAIFiles = NULL)

Arguments
- **BAMFiles**
  - BAMFiles

- **GR**
  - A genomic ranges object that contains the genomic coordinates of the segments.

- **sampleNames**
  - The corresponding sample names to the BAM Files.

- **mode**
  - Possible values are "paired" and "unpaired", whether the mapping algorithm was using a "paired" or "unpaired" strategy.

- **parallel**
  - The number of parallel processes to be used for this function. Default=0.

- **BAIFiles**
  - The names of the BAI files that belong to the BAM files. The vector has to be in the same order as the vector BAMFiles. If the BAI files have the same name as the BAM files, only with ".bai" attached, this parameter needs not be set. (Default = NULL).

Value
An instance of "GRanges", that contains the breakpoints of the initial segments and the raw read counts that were extracted from the BAM files. This object can be used as input for cn.mops and other CNV detection methods.

Author(s)
Guenter Klambauer <klambauer@bioinf.jku.at>
Examples

BAMFiles <- list.files(system.file("extdata", package="cn.mops"), pattern=".bam$", full.names=TRUE)
gr <- GRanges(c("20","20"), IRanges(c(60000,70000), c(70000,80000)))
bamDataRanges <- getSegmentReadCountsFromBAM(BAMFiles, GR=gr, mode="unpaired")
bamDataRanges <- getSegmentReadCountsFromBAM(BAMFiles, GR=gr, mode="unpaired", parallel=2)

This generic function returns the genomic ranges of a CNV detection method stored in an instance of CNVDetectionResult-class.

Description

This generic function returns the genomic ranges of a CNV detection method stored in an instance of CNVDetectionResult-class.

Arguments

object An instance of "CNVDetectionResult".

Value

normalizedData returns a "GRanges" object containing the normalized data.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
gr(r)

This generic function returns the genomic ranges of a CNV detection method stored in an instance of CNVDetectionResult-class.

Usage

## S4 method for signature 'CNVDetectionResult'
gr(object)
haplocn.mops

Arguments

object       An instance of "CNVDetectionResult".

Value

normalizedData returns a "GRanges" object containing the normalized data.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
gr(r)

haplocn.mops

Copy number detection in NGS data of haploid samples.

Description

Performs the cn.mops algorithm for copy number detection in NGS data adjusted to haploid genomes. It is assumed that the normal state is copy number 1. This is an experimental method at the moment.

Usage

haplocn.mops(input, I = c(0.025, 1, 2, 3, 4, 5, 6, 7, 8), classes = c("CN0", "CN1", "CN2", "CN3", "CN4", "CN5", "CN6", "CN7", "CN8"), priorImpact = 1, cyc = 20, parallel = 0, norm = 1, normType = "poisson", sizeFactor = "mean", normQu = 0.25, quSizeFactor = 0.75, upperThreshold = 0.6, lowerThreshold = -0.9, minWidth = 3, segAlgorithm = "fast", minReadCount = 1, returnPosterior = FALSE, ...)

Arguments

input       Either an instance of "GRanges" or a raw data matrix, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region.

I           Vector positive real values that contain the expected fold change of the copy number classes. Length of this vector must be equal to the length of the "classes" parameter vector. For copy number polymorphisms in haploid organisms we suggest to use the default I = c(0.025,1,2,3,4,5,6,7,8).

classes     Vector of characters of the same length as the parameter vector "I". One vector element must be named "CN1". The names reflect the labels of the copy number classes. Default = c("CN0","CN1","CN2","CN3","CN4","CN5","CN6","CN7","CN8").

priorImpact Positive real value that reflects how strong the prior assumption affects the result. The higher the value the more samples will be assumed to have copy number 1. Default = 1.

cyc         Positive integer that sets the number of cycles for the algorithm. Usually after less than 15 cycles convergence is reached. Default = 20.
parallel How many cores are used for the computation. If set to zero than no parallelization is applied. Default = 0.

norm The normalization strategy to be used. If set to 0 the read counts are not normalized and cn.mops does not model different coverages. If set to 1 the read counts are normalized. If set to 2 the read counts are not normalized and cn.mops models different coverages. (Default=1).

normType Mode of the normalization technique. Possible values are "mean", "min", "median", "quant", "poisson" and "mode". Read counts will be scaled sample-wise. Default = "poisson".

sizeFactor By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").

normQu Real value between 0 and 1. If the "normType" parameter is set to "quant" then this parameter sets the quantile that is used for the normalization. Default = 0.25.

quSizeFactor Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization". Real value between 0 and 1. Default = 0.75.

upperThreshold Positive real value that sets the cut-off for copy number gains. All CNV calling values above this value will be called as "gain". The value should be set close to the log2 of the expected foldchange for copy number 3 or 4. Default = 0.5.

lowerThreshold Negative real value that sets the cut-off for copy number losses. All CNV calling values below this value will be called as "loss". The value should be set close to the log2 of the expected foldchange for copy number 1 or 0. Default = -0.9.

minWidth Positive integer that is exactly the parameter "min.width" of the "segment" function of "DNAcopy". minWidth is the minimum number of segments a CNV should span. Default = 4.

segAlgorithm Which segmentation algorithm should be used. If set to "DNAcopy" circular binary segmentation is performed. Any other value will initiate the use of our fast segmentation algorithm. Default = "fast".

minReadCount If all samples are below this value the algorithm will return the prior knowledge. This prevents that the algorithm from being applied to segments with very low coverage.

returnPosterior Flag that decides whether the posterior probabilities should be returned. The posterior probabilities have a dimension of samples times copy number states times genomic regions and therefore consume a lot of memory. Default=FALSE.

Value An instance of "CNVDetectionResult".

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>
individualCall

This generic function returns the individual calls of a CNV detection method stored in an instance of CNVDetectionResult-class.

Description

This generic function returns the individual calls of a CNV detection method stored in an instance of CNVDetectionResult-class.

Arguments

- **object**: An instance of "CNVDetectionResult"

Value

individualCalls returns a "GRanges" object containing the individual calls.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
individualCall(r)
```

individualCall, CNVDetectionResult-method

This generic function returns the individual calls of a CNV detection method stored in an instance of CNVDetectionResult-class.

Description

This generic function returns the individual calls of a CNV detection method stored in an instance of CNVDetectionResult-class.

Usage

```r
## S4 method for signature 'CNVDetectionResult'
individualCall(object)
```

Arguments

- **object**: An instance of "CNVDetectionResult"
iniCall

This generic function returns the informative/non-informative call of a CNV detection method stored in an instance of CNVDetectionResult-class. The I/NI call is a measure for a genomic segment across all samples, whether this segment is a CNV region (informative) or a normal genomic region (non-informative).

Arguments

object An instance of "CNVDetectionResult"

Value

iniCall returns a "GRanges" object containing the individual calls.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
iniCall(r)
This generic function returns the informative/non-informative call of a CNV detection method stored in an instance of `CNVDetectionResult-class`. The I/NI call is a measure for a genomic segment across all samples, whether this segment is a CNV region (informative) or a normal genomic region (non-informative).

Description

This generic function returns the informative/non-informative call of a CNV detection method stored in an instance of `CNVDetectionResult-class`. The I/NI call is a measure for a genomic segment across all samples, whether this segment is a CNV region (informative) or a normal genomic region (non-informative).

Usage

```r
## S4 method for signature 'CNVDetectionResult'
iniCall(object)
```

Arguments

- `object` An instance of "CNVDetectionResult"

Value

`iniCall` returns a "GRanges" object containing the individual calls.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
iniCall(r)
```

This generic function returns the integer copy numbers of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Description

This generic function returns the integer copy numbers of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Arguments

- `object` An instance of "CNVDetectionResult"
Value

integerCopyNumber returns a "GRanges" object containing the integer copy numbers.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
integerCopyNumber(r)
localAssessments

This generic function returns the local assessments, i.e. signed individual informative/non-informative calls, of a CNV detection method stored in an instance of `CNVDetectionResult-class`. For other CNV detection methods this can be (log-) ratios or z-scores.

Description

This generic function returns the local assessments, i.e. signed individual informative/non-informative calls, of a CNV detection method stored in an instance of `CNVDetectionResult-class`. For other CNV detection methods this can be (log-) ratios or z-scores.

Arguments

object

An instance of "CNVDetectionResult"

Value

`localAssessments` returns a "GRanges" object containing the local assessments.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
localAssessments(r)
makeRobustCNVR

Value

localAssessments returns a "GRanges" object containing the local assessments.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
localAssessments(r)

makeRobustCNVR

Calculates robust CNV regions.

Description

This generic function calculates robust CNV regions by segmenting the I/NI call per genomic region of an object CNVDetectionResult-class.

Usage

## S4 method for signature 'CNVDetectionResult'
makeRobustCNVR(object, robust = 0.5,
    minWidth = 4, ...)

Arguments

object An instance of "CNVDetectionResult"

robust Robustness parameter. The higher the value, the more samples are required to have a CNV that confirms the CNV region. Setting this parameter to 0 restores the original CNV regions. (Default=0.5)

minWidth The minimum length measured in genomic regions a CNV region has to span in order to be called. A parameter of the segmentation algorithm. (Default=4).

... Additional parameters passed to the segmentation algorithm.

Details

This generic function calculates robust CNV regions by segmenting the I/NI call per genomic region of an object CNVDetectionResult-class.

cn.mops usually reports a CNV region if at least one individual has a CNV in this region. For some applications it is useful to find more common CNV regions, i.e., regions in which more than one sample has a CNV. The I/NI call measures both signal strength and how many sample show an abnormal copy number, therefore segmentation of the I/NI call can provide robust CNV regions.

Value

makeRobustCNVR returns a "CNVDetectionResult" object containing new values in the slot "cnvr".
normalizeChromosomes

Author(s)
Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
rr <- calcIntegerCopyNumbers(makeRobustCNVR(r, robust=0.1, minWidth=3))
```

normalizeChromosomes  Normalization of NGS data.

Description
Normalize quantitative NGS data in order to make counts comparable over samples, i.e., correcting for different library sizes or coverages. Scales each samples’ reads such that the coverage is even for all samples after normalization.

Usage

```r
normalizeChromosomes(X, chr, normType = "poisson", sizeFactor = "mean", qu = 0.25, quSizeFactor = 0.75, ploidy)
```

Arguments

- **X** Matrix of positive real values, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region. Alternatively this can be a GRanges object containing the read counts as values.
- **chr** Character vector that has as many elements as "X" has rows. The vector assigns each genomic segment to a reference sequence (chromosome).
- **normType** Type of the normalization technique. Each samples’ read counts are scaled such that the total number of reads are comparable across samples. If this parameter is set to the value "mode", the read counts are scaled such that each samples’ most frequent value (the "mode") is equal after normalization. Accordingly for the other options are "mean", "median", "poisson", "quant", and "mode". Default = "poisson".
- **sizeFactor** By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").
- **qu** Quantile of the normType if normType is set to "quant". Real value between 0 and 1. Default = 0.25.
- **quSizeFactor** Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization". Real value between 0 and 1. Default = 0.75.
- **ploidy** An integer value for each sample or each column in the read count matrix. At least two samples must have a ploidy of 2. Default = "missing".

Value
A data matrix of normalized read counts with the same dimensions as the input matrix X.
normalizedData, CNVDetectionResult-method

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
X.norm <- normalizeChromosomes(X)

normalizedData

This generic function returns the normalized data of a CNV detection method stored in an instance of CNVDetectionResult-class.

Description

This generic function returns the normalized data of a CNV detection method stored in an instance of CNVDetectionResult-class.

Arguments

object

An instance of "CNVDetectionResult".

Value

normalizedData returns a "GRanges" object containing the normalized data.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
normalizedData(r)

normalizedData, CNVDetectionResult-method

This generic function returns the normalized data of a CNV detection method stored in an instance of CNVDetectionResult-class.

Description

This generic function returns the normalized data of a CNV detection method stored in an instance of CNVDetectionResult-class.

Usage

## S4 method for signature 'CNVDetectionResult'
normalizedData(object)
**normalizeGenome**

**Arguments**

- **object**: An instance of "CNVDetectionResult".

**Value**

`normalizedData` returns a "GRanges" object containing the normalized data.

**Author(s)**

Guenter Klambauer <klambauer@bioinf.jku.at>

**Examples**

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
normalizedData(r)
```

---

**normalizeGenome**  
*Normalization of NGS data*

**Description**

Normalize quantitative NGS data in order to make counts comparable over samples. Scales each samples' reads such that the coverage is even for all samples after normalization.

**Usage**

```r
normalizeGenome(X, normType = "poisson", sizeFactor = "mean", qu = 0.25,
                 quSizeFactor = 0.75, ploidy)
```

**Arguments**

- **X**: Matrix of positive real values, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region. Alternatively this can be a GRanges object containing the read counts as values.

- **normType**: Type of the normalization technique. Each samples' read counts are scaled such that the total number of reads are comparable across samples. If this parameter is set to the value "mode", the read counts are scaled such that each samples' most frequent value (the "mode") is equal after normalization. Accordingly for the other options are "mean","median","poisson", "quant", and "mode". Default = "poisson".

- **sizeFactor**: By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").

- **qu**: Quantile of the normType if normType is set to "quant". Real value between 0 and 1. Default = 0.25.

- **quSizeFactor**: Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization”. Real value between 0 and 1. Default = 0.75.

- **ploidy**: An integer value for each sample or each column in the read count matrix. At least two samples must have a ploidy of 2. Default = "missing".
Value

A data matrix of normalized read counts with the same dimensions as the input matrix X.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
X.norm <- normalizeGenome(X)

params(r)

This generic function returns the parameters of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Description

This generic function returns the parameters of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Arguments

- object: An instance of "CNVDetectionResult"

Value

params returns a "GRanges" object containing the parameters.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
params(r)
This generic function returns the parameters of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

**Description**

This generic function returns the parameters of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

**Usage**

```r
## S4 method for signature 'CNVDetectionResult'
params(object)
```

**Arguments**

- `object`: An instance of "CNVDetectionResult"

**Value**

`params` returns a `GRanges` object containing the parameters.

**Author(s)**

Guenter Klambauer <klambauer@bioinf.jku.at>

**Examples**

```r
data(cn.mops)
r <- cn.mops(x[1:100,1:5])
params(r)
```

---

**plot**

Plots a `CNVDetectionResult`

**Description**

Plots read counts, call values and CNV calls in an identified CNV region.

**Usage**

```r
## S4 method for signature 'CNVDetectionResult,missing'
plot(x,
     which,margin=c(10,10),toFile=FALSE)
```
posteriorProbs

**Arguments**

- `x` An instance of "CNVDetectionResult"
- `which` The index of the CNV region to be plotted.
- `margin` Vector of two positive integers that states how many segments left and right of the CNV region should be included in the plot. Default = c(10,10).
- `toFile` Logical value whether the output should be plotted to a file. Default = FALSE.

**Value**

Generates a CNV calling plot.

**Author(s)**

Guenter Klambauer <klambauer@bioinf.jku.at>

---

**posteriorProbs**

This generic function returns the posterior probabilities of a CNV detection method stored in an instance of `CNVDetectionResult-class`. The posterior probabilities are represented as a three dimensional array, where the three dimensions are segment, copy number and individual.

**Description**

This generic function returns the posterior probabilities of a CNV detection method stored in an instance of `CNVDetectionResult-class`. The posterior probabilities are represented as a three dimensional array, where the three dimensions are segment, copy number and individual.

**Arguments**

- `object` An instance of "CNVDetectionResult"

**Value**

`posteriorProbs` returns a three dimensional array.

**Author(s)**

Guenter Klambauer <klambauer@bioinf.jku.at>

**Examples**

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
posteriorProbs(r)
```
posteriorProbs,CNVDetectionResult-method

This generic function returns the posterior probabilities of a CNV detection method stored in an instance of `CNVDetectionResult-class`. The posterior probabilities are represented as a three dimensional array, where the three dimensions are segment, copy number and individual.

Description

This generic function returns the posterior probabilities of a CNV detection method stored in an instance of `CNVDetectionResult-class`. The posterior probabilities are represented as a three dimensional array, where the three dimensions are segment, copy number and individual.

Usage

```r
## S4 method for signature 'CNVDetectionResult'
posteriorProbs(object)
```

Arguments

- `object`: An instance of "CNVDetectionResult"

Value

`posteriorProbs` returns a three dimensional array.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

```r
data(cn.mops)
r <- cn.mops(X[1:100,1:5])
posteriorProbs(r)
```

Description

Copy number detection in NGS data with in a control versus cases setting.

This function performs the an alternative version of the cn.mops algorithm adapted to a setting of control versus cases.
Usage

referencecn.mops(cases, controls, I = c(0.025, 0.5, 1, 1.5, 2, 2.5, 3, 3.5, 4, 8, 16, 32, 64), classes = paste("CN", c(0:8, 16, 32, 64, 128), sep = ""), priorImpact = 1, cyc = 20, parallel = 0, norm = 1, normType = "poisson", sizeFactor = "mean", normQu = 0.25, quSizeFactor = 0.75, upperThreshold = 0.5, lowerThreshold = -0.9, minWidth = 4, segAlgorithm = "DNAcopy", minReadCount = 1, verbose = 1, returnPosterior = FALSE, ...)

Arguments

cases Either an instance of "GRanges" or a raw data matrix, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region.

controls Either an instance of "GRanges" or a raw data matrix, where columns are interpreted as samples and rows as genomic regions. An entry is the read count of a sample in the genomic region.

I Vector positive real values that contain the expected fold change of the copy number classes. Length of this vector must be equal to the length of the "classes" parameter vector. For human copy number polymorphisms we suggest to use the default I = c(0.025, 0.5, 1, 1.5, 2, 2.5, 3, 3.5, 4, 8, 16, 32, 64).

classes Vector of characters of the same length as the parameter vector "I". One vector element must be named "CN2". The names reflect the labels of the copy number classes. Default = paste("CN",c(0:8,16,32,64,128),sep="").

priorImpact Positive real value that reflects how strong the prior assumption affects the result. The higher the value the more samples will be assumed to have copy number 2. Default = 1.

cyc Positive integer that sets the number of cycles for the algorithm. Usually after less than 15 cycles convergence is reached. Default = 20.

parallel How many cores are used for the computation. If set to zero than no parallelization is applied. Default = 0.

norm The normalization strategy to be used. If set to 0 the read counts are not normalized and cn.mops does not model different coverages. If set to 1 the read counts are normalized. If set to 2 the read counts are not normalized and cn.mops models different coverages. (Default=1).

normType Mode of the normalization technique. Possible values are "mean","min","median","quant", "poisson" and "mode". Read counts will be scaled sample-wise. Default = "poisson".

sizeFactor By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").

normQu Real value between 0 and 1. If the "normType" parameter is set to "quant" then this parameter sets the quantile that is used for the normalization. Default = 0.25.

quSizeFactor Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization". Real value between 0 and 1. Default = 0.75.

upperThreshold Positive real value that sets the cut-off for copy number gains. All CNV calling values above this value will be called as "gain". The value should be set close to the log2 of the expected foldchange for copy number 3 or 4. Default = 0.5.
sampleNames

lowerThreshold  Negative real value that sets the cut-off for copy number losses. All CNV calling values below this value will be called as "loss". The value should be set close to the log2 of the expected foldchange for copy number 1 or 0. Default = -0.9.

minWidth  Positive integer that is exactly the parameter "min.width" of the "segment" function of "DNAcopy". minWidth is the minimum number of segments a CNV should span. Default = 3.

segAlgorithm  Which segmentation algorithm should be used. If set to "DNAcopy" circular binary segmentation is performed. Any other value will initiate the use of our fast segmentation algorithm. Default = "DNAcopy".

minReadCount  If all samples are below this value the algorithm will return the prior knowledge. This prevents that the algorithm from being applied to segments with very low coverage. Default=1.

verbose  Flag that decides whether referencecn.mops gives status if (verbose>0) messages. Default=1.

returnPosterior  Flag that decides whether the posterior probabilities should be returned. The posterior probabilities have a dimension of samples times copy number states times genomic regions and therefore consume a lot of memory. Default=FALSE.

...  Additional parameters will be passed to the "DNAcopy" or the standard segmentation algorithm.

Value

An instance of "CNVDetectionResult".

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
referencecn.mops(X[1:200, ], apply(X[1:200, ], 1, median))
referencecn.mops(X[1:200, ], apply(X[1:200, ], 1, median), parallel=2)

sampleNames  This generic function returns the sample names of a CNV detection method stored in an instance of CNVDetectionResult-class.

Description

This generic function returns the sample names of a CNV detection method stored in an instance of CNVDetectionResult-class.

Arguments

object  An instance of "CNVDetectionResult"

Value

sampleNames returns a eturns a "GRanges" object containing the parameters.
Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
sampleNames(r)

Description

This generic function returns the sample names of a CNV detection method stored in an instance of `CNVDetectionResult-class`.

Usage

```r
## S4 method for signature 'CNVDetectionResult'
sampleNames(object)
```

Arguments

- `object` An instance of "CNVDetectionResult"

Value

`samleNames` returns a GRanges object containing the parameters.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
sampleNames(r)
**Description**

Performs a fast segmentation algorithm based on the cyber t test and the t statistics. This is a special version for log-ratios or INI calls that are assumed to be centered around 0. For segmentation of data with different characteristics you can a) substract the mean/median/mode from your data or b) use the more general version of this algorithm in the R Bioconductor package "fastseg".

**Usage**

segment(x, alpha = 0.05, segMedianT = NULL, minSeg = 3, eps = 0, delta = 20, maxInt = 40, cyberWeight = 50)

**Arguments**

- **x**: Values to be segmented.
- **alpha**: Real value between 0 and 1 is interpreted as the percentage of total points that are considered as initial breakpoints. An integer greater than 1 is interpreted as number of initial breakpoints. Default = 0.05.
- **segMedianT**: Vector of length 2. Thresholds on the segment’s median. Segments’ medians above the first element are considered as gains and below the second value as losses. If set to NULL the segmentation algorithm tries to determine the thresholds itself. If set to 0 the gain and loss segments are not merged. (Default = NULL).
- **minSeg**: Minimum length of segments. Default = 3.
- **eps**: Real value greater or equal zero. A breakpoint is only possible between to consecutive values of x that have a distance of at least "eps". Default = 0.
- **delta**: Positive integer. A parameter to make the segmentation more efficient. If the statistics of a breakpoint lowers while extending the window, the algorithm extends the windows by "delta" more points until it stops. Default = 20.
- **maxInt**: The maximum length of a segment left of the breakpoint and right of the breakpoint that is considered. Default = 40.
- **cyberWeight**: The "nu" parameter of the cyber t-test. Default = 50.

**Value**

A data frame containing the segments.

**Author(s)**

Guenter Klambauer <klambauer@bioinf.jku.at>

**Examples**

```r
x <- rnorm(n=500,sd=0.5)
x[150:200] <- rnorm(n=51,mean=3,sd=0.5)
segment(x)
```
segmentation, CNVDetectionResult-method

Description

This generic function returns segmentation of a CNV detection method stored in an instance of CNVDetectionResult-class.

Arguments

object

An instance of "CNVDetectionResult"

Value

segmentation returns a "GRanges" object containing the segmentation.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
segmentation(r)
**Examples**

data(cn.mops)
r <- cn.mops(X[1:100,1:5])
segmentation(r)

---

**Description**

Plots the log normalized read counts and the detected segments as a segmentation plot.

**Arguments**

- `r` An instance of "CNVDetectionResult"
- `mainCN` The name of the main copy number. That is "CN2" for diploid individuals. For haplo.cn.mops this should be set to "CN1".
- `sampleIdx` The index of the samples to be plotted. (Default = missing)
- `seqnames` The names of the reference sequence (chromosomes) to be plotted. (Default = missing)
- `segStat` Whether the segment line should display the mean or the median of a segments calls. (Default = "mean").
- `plot.type` The type of plot. (Default = "s").
- `altcol` Logical flag to indicate if chromosomes should be plotted in alternating colors in the whole genome plot. (Default = TRUE).
- `sbyc.layout` Layout settings for the multifigure grid layout for the ‘samplebychrom’ type. It should be specified as a vector of two integers which are the number of rows and columns. The default values are chosen based on the number of chromosomes to produce a near square graph. For normal genome it is 4x6 (24 chromosomes) plotted by rows. (Default = NULL).
- `cbys.layout` Layout settings for the multifigure grid layout for the ‘chrombysample’ type. As above it should be specified as number of rows and columns and the default chosen based on the number of samples. (Default = NULL).
- `cbys.nchrom` The number of chromosomes per page in the layout. (Default = 1).
- `include.means` Logical flag to indicate whether segment means are to be drawn. (Default = TRUE).
- `zeroline` Logical flag to indicate whether a horizontal line at y=0 is to be drawn. (Default = TRUE).
- `pt.pch` The plotting character used for plotting the log-ratio values. (Default = ".")
- `pt.cex` The size of plotting character used for the log-ratio values (Default = 3).
- `pt.cols` The color list for the points. The colors alternate between chromosomes. (Default = c("green","black").)
segplot, CNVDetectionResult-method

Visualization of a CNV detection result.

Description

Plots the log normalized read counts and the detected segments as a segmentation plot.

Usage

```r
## S4 method for signature 'CNVDetectionResult'
segplot(r, mainCN = "CN2", sampleIdx, seqnames,
        segStat = "mean", plot.type = "s", altcol = TRUE, sbyc.layout,
        cbys.nchrom = 1, cbys.layout, include.means = TRUE, zeroline = TRUE,
        pt.pch = ".", pt.cex = 3, pt.cols = c("green", "black"),
        segcol = "red", zlcol = "grey", ylim, lwd = 3, ...)
```

Arguments

- `r`: An instance of "CNVDetectionResult"
- `mainCN`: The name of the main copy number. That is "CN2" for diploid individuals. For haploCN.mops this should be set to "CN1".
- `sampleIdx`: The index of the samples to be plotted. (Default = missing)
- `seqnames`: The names of the reference sequence (chromosomes) to be plotted. (Default = missing)
- `segStat`: Whether the segment line should display the mean or the median of a segments calls. (Default = "mean").
- `plot.type`: the type of plot. (Default = "s").

Value

Generates a segmentation plot.

Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

Examples

```r
data(cn.mops)
r <- cn.mops(X[1:200, ])
segplot(r, sampleIdx=1)
```
altcol logical flag to indicate if chromosomes should be plotted in alternating colors in the whole genome plot. (Default = TRUE).

sbyc.layout layout settings for the multifigure grid layout for the 'samplebychrom' type. It should be specified as a vector of two integers which are the number of rows and columns. The default values are chosen based on the number of chromosomes to produce a near square graph. For normal genome it is 4x6 (24 chromosomes) plotted by rows. (Default = NULL).

cbys.nchrom the number of chromosomes per page in the layout. (Default = 1).

cbys.layout layout settings for the multifigure grid layout for the 'chrombysample' type. As above it should be specified as number of rows and columns and the default chosen based on the number of samples. (Default = NULL).

include.means logical flag to indicate whether segment means are to be drawn. (Default = TRUE).

zeroline logical flag to indicate whether a horizontal line at y=0 is to be drawn. (Default = TRUE).

pt.pch the plotting character used for plotting the log-ratio values. (Default = ".").

pt.cex the size of plotting character used for the log-ratio values (Default = 3).

pt.cols the color list for the points. The colors alternate between chromosomes. (Default = c("green","black").)

segcol the color of the lines indicating the segment means. (Default = "red").

z1col the color of the zeroline. (Default = "grey").

ylim this argument is present to override the default limits which is the range of symmetrized log-ratios. (Default = NULL).

lwd line weight of lines for segment mean and zeroline. (Default = 3).

... other arguments which will be passed to plot commands.

Value
Generates a segmentation plot.

Author(s)
Guenter Klambauer <klambauer@bioinf.jku.at>

Examples
data(cn.mops)
r <- cn.mops(X[1:200, ])
segplot(r,sampleIdx=1)
### show

Displays the result object of a copy number detection method.

#### Description

Displays method for S4 class `CNVDetectionResult`.

#### Usage

```r
## S4 method for signature 'CNVDetectionResult'
show(object)
```

#### Arguments

- `object`: An instance of a "CNVDetectionResult".

#### Value

Displays the result object of a CNV detection method.

#### Author(s)

Guenter Klambauer <klambauer@bioinf.jku.at>

---

### singlecn.mops

Copy number detection in NGS data with in a setting in which only one sample is available

#### Description

This function performs the an alternative version of the cn.mops algorithm adapted to a setting of a single sample.

#### Usage

```r
singlecn.mops(x, I = c(0.025, 0.5, 1, 1.5, 2, 2.5, 3, 3.5, 4),
   classes = c("CN0", "CN1", "CN2", "CN3", "CN4", "CN5", "CN6", "CN7", "CN8"),
   priorImpact = 1, cyc = 20, parallel = 0, norm = 1,
   normType = "poisson", sizeFactor = "mean", normQu = 0.25,
   quSizeFactor = 0.75, upperThreshold = 0.5, lowerThreshold = -0.9,
   minWidth = 3, segAlgorithm = "fast", minReadCount = 1,
   returnPosterior = FALSE, ...)
```
Arguments

- **x**: Either an instance of "GRanges" or a raw data matrix with one column or a vector of read counts. An entry is the read count of the sample in the genomic region.

- **I**: Vector positive real values that contain the expected fold change of the copy number classes. Length of this vector must be equal to the length of the "classes" parameter vector. For human copy number polymorphisms we suggest to use the default \( I = c(0.025,0.5,1,1.5,2,2.5,3,3.5,4) \).

- **classes**: Vector of characters of the same length as the parameter vector "I". One vector element must be named "CN2". The names reflect the labels of the copy number classes. Default = c("CN0","CN1","CN2","CN3","CN4","CN5","CN6","CN7","CN8").

- **priorImpact**: Positive real value that reflects how strong the prior assumption affects the result. The higher the value the more samples will be assumed to have copy number 2. Default = 1.

- **cyc**: Positive integer that sets the number of cycles for the algorithm. Usually after less than 15 cycles convergence is reached. Default = 20.

- **parallel**: How many cores are used for the computation. If set to zero than no parallelization is applied. Default = 0.

- **norm**: The normalization strategy to be used. If set to 0 the read counts are not normalized and cn.mops does not model different coverages. If set to 1 the read counts are normalized. If set to 2 the read counts are not normalized and cn.mops models different coverages. (Default=1).

- **normType**: Mode of the normalization technique. Possible values are "mean","min","median","quant", "poisson" and "mode". Read counts will be scaled sample-wise. Default = "poisson".

- **sizeFactor**: By this parameter one can decide to how the size factors are calculated. Possible choices are the the mean, median or mode coverage ("mean", "median", "mode") or any quantile ("quant").

- **normQu**: Real value between 0 and 1. If the "normType" parameter is set to "quant" then this parameter sets the quantile that is used for the normalization. Default = 0.25.

- **quSizeFactor**: Quantile of the sizeFactor if sizeFactor is set to "quant". 0.75 corresponds to "upper quartile normalization". Real value between 0 and 1. Default = 0.75.

- **upperThreshold**: Positive real value that sets the cut-off for copy number gains. All CNV calling values above this value will be called as "gain". The value should be set close to the \( \log2 \) of the expected foldchange for copy number 3 or 4. Default = 0.5.

- **lowerThreshold**: Negative real value that sets the cut-off for copy number losses. All CNV calling values below this value will be called as "loss". The value should be set close to the \( \log2 \) of the expected foldchange for copy number 1 or 0. Default = -0.9.

- **minWidth**: Positive integer that is exactly the parameter "min.width" of the "segment" function of "DNAcopy". minWidth is the minimum number of segments a CNV should span. Default = 3.

- **segAlgorithm**: Which segmentation algorithm should be used. If set to "DNAcopy" circular binary segmentation is performed. Any other value will initiate the use of our fast segmentation algorithm. Default = "fast".

- **minReadCount**: If all samples are below this value the algorithm will return the prior knowledge. This prevents that the algorithm from being applied to segments with very low coverage. Default=1.
returnPosterior

Flag that decides whether the posterior probabilities should be returned. The posterior probabilities have a dimension of samples times copy number states times genomic regions and therefore consume a lot of memory. Default=FALSE.

Additional parameters will be passed to the "DNAcopy" or the standard segmentation algorithm.

Value

An instance of "CNVDetectionResult".

Author(s)

Guenther Klambauer <klambauer@bioinf.jku.at>

Examples

data(cn.mops)
singlecn.mops(XRanges[,1])

Description

This data set gives the read counts of 40 samples in 5000 genomic locations. The rows correspond to genomic segments of 25kbp length and the columns to samples. An entry is the number of reads that map to the specific segment of the sample. The rownames contain the information of the genomic location - they are in the format refseqname_startposition_endposition. The simulated data contains CNVs given in the CNVRanges object. It was generated using distributions of read counts as they appear in real sequencing experiments. CNVs were implanted under the assumption that the expected read count is linear dependent on the copy number (e.g. in a certain genomic we expect $\lambda$ reads for copy number 2, then we expect $2 \cdot \lambda$ reads for copy number 4).

Usage

X

Format

A data matrix of 5000 rows and 40 columns.

Source


References

XRanges

A simulated data set for CNV detection from NGS data.

Description

This data set gives the read counts of 40 samples in 5000 genomic locations. The rows correspond to genomic segments of 25kbp length and the columns to samples. An entry is the number of reads that map to the specific segment of the sample. The "GRanges" object contains the name of the reference sequence, start and end position of the genomic segments. The simulated data contains CNVs given in the CNVRanges object. It was generated using distributions of read counts as they appear in real sequencing experiments. CNVs were implanted under the assumption that the expected read count is linear dependent on the copy number (e.g. in a certain genomic we expect \( \lambda \) reads for copy number 2, then we expect \( 2 \cdot \lambda \) reads for copy number 4).

Usage

XRanges

Format

A GRanges object with 5000 rows and 40 value columns across 1 space.

Source


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