## Package 'seqCBS'

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Type Package

Title Copy Number Profiling using Sequencing and CBS

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**Description** This is a method for DNA Copy Number Profiling using Next-Generation Sequencing. It has new model and test statistics based on non-homogeneous Poisson Processes with change point models. It uses an adaptation of Circular Binary Segmentation. Also included are methods for point-wise Bayesian Confidence Interval and model selection method for the change-point model. A case and a control sample reads (normal and tumor) are required.

License GPL-2

LazyLoad yes

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seqCBS-package

Scan Statistics CNV detection using sequencing data

## Description

CNV detection using matched case-control sequencing read data. It gives a number of scan statistics for the detection of rate differences between two non-homogeneous Poisson Processes, and modified BIC model selection.

## Details

Package:	seqCBS
Type:	Package
Version:	1.2
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License:	GPL-2
LazyLoad:	yes

~~ An overview of how to use the package, including the most important ~~ ~~ functions ~~

## **BayesCptCI**

## Author(s)

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BayesCptCI

Bayesian Point-wise Confidence Interval for Change-Point Model

## Description

This algorithm computes a point-wise Bayesian CI for the p parameter in change-point model on binomial process.

#### Usage

## Arguments

cases	A numeric vector of the case/tumor reads
controls	A numeric vector of the control/normal reads
CBSRes	Output from the ScanCBS algorithm on this case/control data
stepSize	An actual point-wise computation is time-consuming; by using stepSize = n, a Bayesian CI is computed at every n reads. The adaptive option gives good computational speed by choosing stepSize based on the data.
adaptMaxMix	An upper bound for the number of unique weights calculated at each change point under the adaptive method. The default is 80 for an average of approximately 5000 mixture components at each point.
alpha	Defaults to 0.05 for the usual CI.
epsilon	The cutoff for the likelihood ratio of a model with shifted change point compared to the ScanCBS estimated change-point. The likelihood decreases exponentially around the true point or a 'good' estimate of it. Only alternatives with above the cutoff likelihood ratio are considered plausible and integrated in following computations.
epsCDF	This is an error tolerance value for finding the quantile of the posterior, which is a Beta mixture distribution.
verbose	If TRUE, then will print much information on each segmentation. For diagnostics only.

#### Details

This method is a Bayesian point-wise CI for our change-point method. It takes model complexity (number of change points) as a given. With the ScanCBS-estimated change points, it evaluates alternatives for the change points around the estimated value and computes the likelihood of the alternative models. Through theoretical derivation, we then have the estimated probability of a case read, p, at a given read index, to have a posterior density given by Beta Mixture. We then compute the quantiles of this distribution using a safe version of Newton-Raphson implemented in C as the CI at this read.

#### Value

CIRes	A matrix containing the location and its CI of p, each column is a location, or a strech of location if stepSize>1
wkRes	The likelihood ratio for alternatives around each estimated change point
mixStruct	The Beta mixture components for each unique location, containing a collection of two shape parameters and the weight wk
timeCIRes	A list containing the result of the timing of this algorithm

## Author(s)

Jeremy J. Shen

#### See Also

ScanBIC

CombineCaseControlC Combine case and control reads

#### Description

Combine the case and control reads; finds the unique read positions and count the number of case and control reads.

## Usage

```
CombineCaseControlC(cases, controls)
```

#### Arguments

cases	A vector of numeric read positions from case sample
controls	A vector of numeric read positions from control sample

## Details

A few C functions are used for efficient implementation

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#### CombineReadsAcrossRuns

## Value

combX	Number of total reads at read position
combZ	Number of case reads at read position
combL	Vector of unique read positions

## Author(s)

Jeremy J. Shen

#### See Also

ScanCBS

CombineReadsAcrossRuns

Combine multiple read lists

## Description

Combines multiple lists in the same format of the same sample into one list of the said format.

#### Usage

```
CombineReadsAcrossRuns(seqs)
```

#### Arguments

seqs A list of lists, each containing equal number of numeric vectors that can be concatenated together. Both number of lists and number of variables can be arbitrary.

## Value

Returns a list of the same format as the input lists

#### Author(s)

Jeremy J. Shen

## See Also

ScanCBS

## Examples

```
data(JSSim_NormalSim1)
data(JSSim_NormalSim2)
write.table(JSSim_NormalSim1, file="JSSim_NormalSim1.txt",
sep="\t", quote=FALSE, row.names=FALSE, col.names=FALSE)
write.table(JSSim_NormalSim2, file="JSSim_NormalSim2.txt",
sep="\t", quote=FALSE, row.names=FALSE, col.names=FALSE)
JSSim_Normal1 = readSeqChiang("JSSim_NormalSim1.txt")
JSSim_Normal2 = readSeqChiang("JSSim_NormalSim2.txt")
file.remove(c("JSSim_NormalSim1.txt", "JSSim_NormalSim2.txt"))
combJSNormal = CombineReadsAcrossRuns(list(JSSim_Normal1, JSSim_Normal2))
print(c(length(JSSim_Normal1$seqF), length(JSSim_Normal2$seqF),
length(combJSNormal$seqF)))
```

getAutoGridSize Get Automatic Grid Sizes

#### Description

This produces a default set of grid sizes to be used in Interative Grid Scan

#### Usage

getAutoGridSize(nL)

#### Arguments

nL Number of unique read positions

#### Details

The default grid sizes are powers of 10

#### Value

numeric vector of grid sizes

#### Author(s)

Jeremy J. Shen

#### See Also

ScanCBS

#### Examples

## Should produce a vector of power of ten up to 10000
getAutoGridSize(2\*10^5)

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getCountsInWindow Get number of reads in fixed-width window

## Description

Computes the number of reads for each fixed-width window between two limits

## Usage

```
getCountsInWindow(events, startE, endE, windowSize = 10000, sorted = FALSE)
```

## Arguments

A vector of the read positions
Left limit
Right Limit
Size of the window
Whether events is sorted, default F

## Details

Uses hist() function

## Value

A vector of counts for each window

## Author(s)

Jeremy J. Shen

## See Also

ScanCBS

## Examples

getCountsInWindow(sample(1:10000, 3000, replace=TRUE), 0, 10000, 100, FALSE)

hppSimulate

#### Description

Simulation of a homogeneous poisson process using poisson and uniform distribution

## Usage

```
hppSimulate(lambda, maxVal)
```

## Arguments

lambda	The rate of the poisson process
maxVal	The maximum length of the process to be observed

## Details

This is a very simple simulation function meant to be used in the NHPP generation.

## Value

Returns a vector of point events generated for this process

## Author(s)

Jeremy J. Shen

#### See Also

nhppSimulate

JSSim\_Meta

Meta File for Simulated Datasets

## Description

This data set contains the links and description for the normal and tumor simulated data sets

#### Usage

JSSim\_Meta

#### Format

A matrix with 3 columns and 4 rows, and a header line

JSSim\_NormalSim1 Simulated normal sample dataset 1

#### Description

This data set contains simulated reads of a truncated chromosome from a normal sample

## Usage

JSSim\_NormalSim1

## Format

A matrix with 3 columns and 15193 rows

JSSim\_NormalSim2 Simulated normal sample dataset 2

#### Description

This data set contains a second set of simulated reads of a truncated chromosome from a normal sample

#### Usage

JSSim\_NormalSim2

## Format

A matrix with 3 columns and 15206 rows

JSSim\_SpikeMat True Signal Spike for the Simulated Dataset

## Description

This data set gives the true signal spike location and strength for the simulated datasets

#### Usage

```
JSSim_SpikeMat
```

## Format

A matrix with 5 columns and rows

JSSim\_TumorSim1

#### Description

This data set contains simulated reads of a truncated chromosome from a Tumor sample, with spiked in signals

#### Usage

JSSim\_TumorSim1

#### Format

A matrix with 3 columns and 16452 rows

JSSim\_TumorSim2 Simulated Tumor sample dataset 2

### Description

This data set contains a second set of simulated reads of a truncated chromosome from a Tumor sample, with spiked in signals

#### Usage

JSSim\_TumorSim2

#### Format

A matrix with 3 columns and 16225 rows

nhppRateEstimate Estimate the rate of non-homogeneous PP with data

## Description

Given a vector of point events, give a rough estimate of the rate of underlying non-homogeneous Poisson process by window and smoothing

#### Usage

```
nhppRateEstimate(controls, length.out = floor(length(controls)/20),
lowessF = 0.1)
```

#### Arguments

controls	A vector of point locations (read positions) of a control sample for which the rate is wanted
length.out	The number of windows to be used for the rate estimate vector; default to be number of observations/100
lowessF	Smoothing factor for the lowess smoothing of the windowed rates, describes the proportion of windows around a particular window that has influence on its smoothed rate estimate

#### Details

This is used to give a realistic estimate of the rate nhpp of control samples

## Value

Returns a vector of length length.out that contains the smoothed rate estimate of each window

#### Author(s)

Jeremy J. Shen

#### See Also

nhppSimulate

```
nhppSimConstWindowAnalysis
```

Analyze the performance on simulation with constant signal length in each set

#### Description

Takes the dataset and metafile output of nhppSimConstWindowGen and of SegSeq, then evaluates the performance in change-point precision and recall. The dataset must be generated in such format for this function to work.

#### Usage

```
nhppSimConstWindowAnalysis(filePrefix, chromosomeN,
    distMetric=c(20,50,100,150,200,300,500,1000),
    cptLen=c(3,5,8,12,15,20,30,50,100),
    nPair=2, nRepeat=10, statistic="normal", grid.size="auto", takeN=5,
    maxNCut=60, minStat=5, verbose=FALSE, timing=TRUE, hasRun=FALSE,
    width=12, height=6)
```

## Arguments

filePrefix	The first part of the filename for data and metafile generated by nhppSimConstWindowGen
chromosomeN	The number indicating the chromosome number the dataset emulates
distMetric	A set of criterions of determining change points called are true. A call is deemed true if an actual signal change points within x number of reads is matched to it, after a minimum-cost bipartite matching. Larger value is a looser criterion.
cptLen	The second part of the filename for data and metafile generated by nhppSimConstWindowGen, indicating the length of the true signal. Constant width of the signal (CN gain or loss) region to simulate, can be a vector of different values for which to test
nPair	A part of the filename for data and metafile generated by nhppSimConstWindowGen, indicating the number of normal/tumor pair. Number of tumor samples to gen- erate for each choice of the width of the signal; number of normal samples to generate
nRepeat	A part of the filename for data and metafile generated by nhppSimConstWindowGen. Number of times to repeat the simulation data generation
statistic	The type of statistic to use for the analysis
grid.size	Argument to ScanCBS
takeN	Argument to ScanCBS
maxNCut	Argument to ScanCBS
minStat	Argument to ScanCBS
verbose	If TRUE, will print run information as the algorithm proceeds
timing	Performs timing of the ScanCBS algorithm
hasRun	If TRUE, will read the output file of ScanCBS instead of run it on these datasets again. Only use when the same call to ScanCBS has been used before in this function call.
width	Width of the graph output file
height	Height of the graph output file

## Details

This function is used in conjunction with nhppSimConstWindowGen. It reads in the data and metafile output of the said function, and compares the performance of our algorithm with SegSeq. It is important that SegSeq has been used on the simulation datasets generated before using this.

#### Value

simCBS	Result of ScanCBS output structure	
CBSMatchDist	The distance among reads after minimum-cost bipartite graph matching for our algorithm	
SegMatchDist	The distance among reads after minimum-cost bipartite graph matching for SegSeq	
CBSRecall, SegRecall		
	The recall rates of two algorithms	

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## nhppSimConstWindowGen

CBSPrecision, SegPrecision		
The precision rates of two algorithms		
CBSFMeasure, SegFMeasure		
The F-measure of two algorithms		
trueTauMeanSigLen		
The mean distance between true signal boundaries		
nTrueTau The number of true change points		
nCBSCall, nSegCall		
Number of change points called by the two algorithms		
CBSTime Mean computational time of ScanCBS for each signal length		

#### Author(s)

Jeremy J. Shen

## See Also

nhppSimConstWindowGen

nhppSimConstWindowGen Simulate a Non-Homogeneous PP with constant window spike

## Description

Simulate non-homogeneous Poisson processes with a number of constant-widths windows of signal spike, and output the data and meta file

### Usage

```
nhppSimConstWindowGen(controlRates, filename, chromosomeN, nSpike=25,
cptLen=c(3,5,8,12,20,30,50,75,100), nPair=2, nRepeat=10, minGain=1.5,
maxGain=4, minLoss=0.01, maxLoss=0.5, pGain=0.6)
```

## Arguments

controlRates	The estimated rate of nhpp for the control
filename	The prefix of all the output files from this simulation
chromosomeN	The chromosome number. Should be the number from which the samples are emulated
nSpike	Number of signal spikes
cptLen	Constant width of the signal (CN gain or loss) region to simulate, can be a vector of different values for which to test
nPair	Number of tumor samples to generate for each choice of the width of the signal; number of normal samples to generate
nRepeat	Number of times to repeat the simulation data generation

minGain	Minimal signal gain
maxGain	Maximal signal gain
minLoss	Minimal signal loss
maxLoss	Maximal signal loss
pGain	Proportion of the signal regions that are CN gain

## Details

This function is used in conjunction with a modified, windowed rate vector to simulate non-homogeneous Poisson processes with a number of constant-widths windows of signal spike. One should use the nhppRateEstimate function to estimate the rate of a control sample one wishes to mimic. This function randomly choose windows of a specified constant width, and spike in signals (change points) which can be either gain or loss of copy numbers.

#### Value

No return value. Generates a number of .txt files, one for each normal/tumor sample as raw data, one input meta file and a file with the true change points for each choice of cptLen.

#### Author(s)

Jeremy J. Shen

#### See Also

nhppSimulate

nhppSimulate	Simulate a non-homogeneous Poisson Process
--------------	--

#### Description

This function simulates an NHPP by blocked thinning

#### Usage

```
nhppSimulate(smoothRates)
```

#### Arguments

smoothRates A list containing x and y, which are the mid-points of the window and the smoothed number of events in this window

#### Details

The list component y of the argument represents the smoothed number of events in the window, namely, they represent the window rate

## nhppSpike

## Value

Returns a vector of events of a realization of the NHPP

#### Author(s)

Jeremy J. Shen

## See Also

nhppRateEstimate, nhppSpike

nhppSpike

Spike rates of NHPP

## Description

Randomly spike the smoothed control rate of an NHPP according to the parameters.

#### Usage

```
nhppSpike(smoothRates, nSpike = 25, cptLenR = 4, cptLenMean = 10,
minGain = 1.5, maxGain = 10, minLoss = 0.01, maxLoss = 0.5, pGain = 0.6)
```

#### Arguments

smoothRates	The smoothed rate estimate of the control process
nSpike	Number of signal spikes
cptLenR	Parameter for signal width (parameter R of negative binomial)
cptLenMean	Parameter for signal width (mean width)
minGain	Minimal Gain relative CN
maxGain	Maximal Gain relative CN
minLoss	Minimal Loss relative CN
maxLoss	Maximal Loss relative CN
pGain	Proportion of signal regions that are CN gain.

## Details

The signal width is randomly generated by negative binomial distribution with the two parameters given. The signal strength are uniformly drawn between the two limits.

## Value

spikeMat	A matrix containing the actual signal spike information
caseRates	The rate of the case PP to be simulated after signal spike

#### Author(s)

Jeremy J. Shen

## See Also

nhppSimulate

nhppSpikeConstWindow Spike NHPP rate with constant window width

#### Description

Randomly spike the smoothed control rate of an NHPP according to the parameters, with constant window width.

#### Usage

```
nhppSpikeConstWindow(smoothRates, nSpike = 25, cptLen = 5, minGain = 1.5,
maxGain = 10, minLoss = 0.01, maxLoss = 0.5, pGain = 0.6)
```

## Arguments

smoothRates	The smoothed rate estimate of the control process
nSpike	Number of signal spikes
cptLen	Window width of each signal region
minGain	Minimal Gain relative CN
maxGain	Maximal Gain relative CN
minLoss	Minimal Loss relative CN
maxLoss	Maximal Loss relative CN
pGain	Proportion of signal regions that are CN gain

#### Details

The signal strength are uniformly drawn between the two limits.

### Value

spikeMat	A matrix containing the actual signal spike information
caseRates	The rate of the case PP to be simulated after signal spike

#### Author(s)

Jeremy J. Shen

## See Also

nhppSimulate

readInput

## Description

This is used to control the read of a meta file containing names of data files, merge, and give usable output for the main program

#### Usage

```
readInput(inputFilename, formatName="Chiang", sep = "\t")
```

## Arguments

inputFilename	The name of file, containing relevant information of all input files
formatName	The format in which the data files are written in. We use the simple 'Chiang' as default format of input.
sep	Delimiter of the meta input file, default is tab-delimited

## Details

The meta input file should be organized in a table format with 2 columns, one of which is 'file' and the other is 'type', indicating the data file names and whether the data is from normal or tumor. We recommend using the 'Chiang' format, as used by the datasets of Chiang (2009). This format requires minimal memory and contains all relevant information for this program. It is a table with two columns, first being the chromosome of the mapped read, and the second being the position of the read in the chromosome. One line for each observation.

#### Value

normalSeq	A list containing the combined normal/control reads
tumorSeq	A list containing the combined case/tumor reads

## Author(s)

Jeremy J. Shen

#### See Also

readListInputFile, readSeq

## Examples

```
# This shows the format of the meta file
data(JSSim_Meta)
print(JSSim_Meta)
# This shows the recommended format, the Chiang data format
data(JSSim_NormalSim1)
print(head(JSSim_NormalSim1))
```

readListInputFile Read meta file containing list of raw data files

## Description

Reads a meta file that contains the file names and type of the data files. See details for the format.

#### Usage

```
readListInputFile(inputFilename, sep = "\t")
```

#### Arguments

inputFilename	The name of file, containing relevant information of all input files
sep	Delimiter of the meta input file, default is tab-delimited

## Details

The meta input file should be organized in a table format with 2 columns, one of which is 'file' and the other is 'type', indicating the data file names and whether the data is from 'normal' or 'tumor'.

#### Value

normalFiles	A character vector containing the names of files with the normal reads
tumorFiles	A character vector containing the names of files with the tumor reads

#### Author(s)

Jeremy J. Shen

#### See Also

readInput, readSeq

#### Examples

```
# This shows the format of the meta file
data(JSSim_Meta)
print(JSSim_Meta)
```

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readSeq

#### Description

This is a wrapper function. It calls one of the subroutines to reads in a datafile, depending on the format

## Usage

readSeq(filename, formatName)

## Arguments

filename	The file name of the data file to be read
formatName	The format the file is in. Can be either 'Chiang' or 'ELANDPaired'. We recom-
	mend using Chiang since this is the minimal required format.

## Details

We recommend using the 'Chiang' format, as used by the datasets of Chiang (2009). This format requires minimal memory and contains all relevant information for this program. It is a table with two columns, first being the chromosome of the mapped read, and the second being the position of the read in the chromosome. One line for each observation. If one has paired read, please use only one of the reads and the mapped location should be the 5'-end.

#### Value

seqF	Read position for each read
seqChr	Chromosome of each mapped read

#### Author(s)

Jeremy J. Shen

#### References

Chiang et al., Nature Methods, 2009, Vol.6 No.1

## See Also

readSeq, readSeqChiang, readSeqELANDPaired

#### Examples

```
# This shows the recommended format, the Chiang data format
data(JSSim_NormalSim1)
print(head(JSSim_NormalSim1))
```

readSeqChiang

## Description

Read data formatted as in Chiang (2009), which we recommend using.

#### Usage

```
readSeqChiang(filename)
```

## Arguments

filename The file name of the data set

#### Details

This format requires minimal memory and contains all relevant information for this program. It is a table with two columns, first being the chromosome of the mapped read, and the second being the position of the read in the chromosome. One line for each observation. In case of paired read, we only use the front read (whichever has a smaller position label) and ask that you use only that for input.

#### Value

seqF	Read position for each read
seqChr	Chromosome of each mapped read

## Author(s)

Jeremy J. Shen

#### References

Chiang et al., Nature Methods, 2009, Vol.6 No.1

#### See Also

readSeq, readSeqChiang, readSeqELANDPaired

## Examples

```
# This shows the format of this type of data
data(JSSim_NormalSim1)
print(head(JSSim_NormalSim1))
```

readSeqELANDPaired Read raw data formatted as in paired ELAND output

#### Description

Read datasets with paired-end format, possible output format of ELAND

#### Usage

readSeqELANDPaired(filename)

## Arguments

filename The file name of the data set

#### Details

This format has two reads per line, each looking like "NACGATGAAACCCCGTCTCTACTAAC-CATACAAAAA hs\_ref\_chr17.fa 12091150 R TGTCGCCCAGGCTGCAATGCAGTGGCGCGATCTCGG hs\_ref\_chr17.fa 12091018 F". There are 8 columns, 4 for each of the paired read. The first is the actual read sequence, which we discard; the second is the chromosome of the mapped read; the third is the read position; and the last is indicating whether it is a front- or rear- end read. We only use the reads with the same mapped chromosome and only the front read. This contains more information than needed; the Chiang format is prefered.

## Value

seqF	Read position for each read
seqChr	Chromosome of each mapped read

## Author(s)

Jeremy J. Shen

#### See Also

readSeq, readSeqChiang

relCNComp

## Description

This computes the relative copy number by each of the segment called

#### Usage

relCNComp(combX, combZ, tauHatInd, p, alpha)

## Arguments

combX	The number of reads at each unique read position
combZ	The number of case/tumor reads at each unique read position
tauHatInd	The index of change points called
р	The overall proportion of case reads
alpha	Significance level for testing whether each segment is a gain (relative $CN > 1$ ) or loss (relative $CN < 1$ ). The method internally corrects for multiple testing.

#### Details

The relative CN is defined as the number of case reads divided by the number of control reads in a window, adjusted for overall proportion of case reads (divided by the overall relative CN).

#### Value

Returns a vector of relative CN for each of the segment between two change points

#### Author(s)

Jeremy J. Shen

ScanBIC

Compute the modified BIC for change-point models

## Description

This computes mBIC for the current change point model. We then use this to determine the appropriate model complexity.

#### Usage

ScanBIC(combX, combZ, tauHat, lik0, nTotal)

## ScanCBS

#### Arguments

combX	The number of reads at each unique read position
combZ	The number of case/tumor reads at each unique read position
tauHat	The change points called
lik0	The null likelihood. Computed in the main routine.
nTotal	The total number of reads

## Details

This is meanted to be called as a subrountine of ScanCBS

## Value

Returns a numerical value of mBIC for the current model

## Author(s)

Jeremy J. Shen

## See Also

ScanCBS

ScanCBS

Main CBS Algorithm for Change-Point Detection

## Description

This is the main algorithm. It teratively scans for window of arbitrary size where the case and control read depths are different. It continues until a stopping criterion based on mBIC, maximum number of cut, and the statistic at the current segment.

## Usage

```
ScanCBS(cases, controls, statistic = "binomial", grid.size = "auto", takeN = 5,
maxNCut = 100, minStat = 0, alpha=0.05, verbose = FALSE, timing = TRUE)
```

#### Arguments

cases	A numeric vector of the case/tumor reads
controls	A numeric vector of the control/normal reads
statistic	The statistic to be used. Can be 'binomial', 'rabinowitz' or 'normal'.
grid.size	The set of grid sizes for the iterative search. An automatic default can be com- puted.

takeN	The number of candidate change points to be added to a temporary set at each grid size
maxNCut	The maximum number of segmentation steps to perform
minStat	The minimum statistic value required to continue the segmentation. Default 0 as this criterion being ignored.
alpha	Significance level for testing whether each segment is a gain (relative $CN > 1$ ) or loss (relative $CN < 1$ ). The method internally corrects for multiple testing.
verbose	If TRUE, then will print much information on each segmentation. For diagnostics only.
timing	If TRUE, perform a timing of this algorithm, include in the output data file.

## Details

This algorithm is an use of the Circular Binary Segmentation method. It continues to segment the reads and consider the resulting child regions for further segmentation. It keeps track of the most promising cut in each children, and only the child region with the most significant segmentation is further cut, yielding more children. This is repeated until stopping criteria are met. The three types of statistics are by the use of exact binomial likelihood ('binomial'), score statistic ('rabinowitz') or using normal approximation to the binomial ('normal').

#### Value

tauHat	The change points called
statHat	A matrix containing the statistic and its segmentation for the model called, in the order of the segmentation. The columns are break points in genomic scale $(1,2)$ , read index scale $(3,4)$ , value of test statistic $(5)$ , the parent segment in genomic scale $(6,7)$ , and mBIC of the model $(8)$ .
relCN	The relative CN computed for each segment between change points
relGainLoss	Test result of whether each segment is a gain, loss, or normal
timingRes	A list containing the result of the timing of this algorithm

#### Author(s)

Jeremy J. Shen

#### References

D. Rabinowitz, IMS Lecture Notes - Monograph Series, Vol. 23, 1994

## See Also

ScanIterateGrid, ScanBIC, relCNComp, getAutoGridSize

ScanCBSPlot

## Description

This is an overall plotting function to display the segmentation for a chromosome

#### Usage

```
ScanCBSPlot(cases, controls, CBSObj, filename, mainTitle, CIObj=NULL,
length.out=10000, localWindow=0.5*10^5, localSeparatePlot=TRUE,
smoothF=0.025, xlabScale=10^6, width=12, height=18)
```

## Arguments

cases	The case read positions (should be restricted to a chromosome)	
controls	The control read positions (should be restricted to a chromosome)	
CBSObj	The output object of the ScanCBS function	
filename	The output file names of the plot	
mainTitle	The title of the plot	
CIObj	Optional; the Bayesian CI computed by BayesCptCI function	
length.out	The number of windows to use for the display of smoothed rate estimates	
localWindow	The number of genome locations to show around each of the called change points	
localSeparatePlot		
	Whether to show the local behavior of each change point in a seperate PDF file. Default to TRUE. The output file are the given filename attached with the index and actual location of the change point.	
smoothF	The lowess smoothing factor. The proportion of windows around the current window that affects its smoothed rate estimate	
xlabScale	The scaling factor of the read positions, often in 10 <sup>6</sup> , or Mb	
width	The width of the output graph in inches	
height	The height of the output graph in inches	

#### Details

This function produces three sub-graphs, showing the segmentation calls, the smoothed rate estimate, and the inferred relative copy number. It is crucial that one seperates the plot for each chromosome. It also makes a zoom-in plot for a region around each of the called change points.

## Value

No return object

## Author(s)

Jeremy J. shen

## See Also

ScanCBS, ScanCBSSimPlot, relCNComp

ScanCBSSimPlot Plotting for CBS results of Simulated Data

#### Description

This is an overall plotting function to display the segmentation for a chromosome, for simulation data.

#### Usage

```
ScanCBSSimPlot(cases, controls, CBSObj, trueTau, SpikeMat, filename, mainTitle,
CIObj=NULL, length.out=10000, localWindow=0.5*10^5, localSeparatePlot=TRUE,
smoothF=0.025, xlabScale=10^6, width=12, height=18)
```

## Arguments

cases	The case read positions (should be restricted to a chromosome)	
controls	The control read positions (should be restricted to a chromosome)	
CBSObj	The output object of the ScanCBS function	
trueTau	The true location of the change points in simulation	
SpikeMat	The matrix of signal spikes as generated by the relevant simulation functions	
filename	The output file names of the plot	
mainTitle	The title of the plot	
CIObj	Optional; the Bayesian CI computed by BayesCptCI function	
length.out	The number of windows to use for the display of smoothed rate estimates	
localWindow	The number of genome locations to show around each of the called change points	
localSeparatePlot		
	Whether to show the local behavior of each change point in a seperate PDF file. Default to TRUE. The output file are the given filename attached with the index and actual location of the change point.	
smoothF	The lowess smoothing factor. The proportion of windows around the current window that affects its smoothed rate estimate	
xlabScale	The scaling factor of the read positions, often in 10 <sup>6</sup> , or Mb	
width	The width of the output graph in inches	
height	The height of the output graph in inches	

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## ScanIterateGrid

## Details

This is similar to ScanCBSPlot. This function produces three sub-graphs, showing the segmentation calls, the smoothed rate estimate, and the inferred relative copy number. It is crucial that one seperates the plot for each chromosome. This also has an option of showing each change point details in seperate graphs.

## Value

No return object

## Author(s)

Jeremy J. Shen

## See Also

ScanCBS, ScanCBSPlot, relCNComp

ScanIterateGrid Main Scan with Iterative Grid Search

## Description

This is a computational speed-up to prevent a quadratic order computation.

## Usage

```
ScanIterateGrid(combX, combZ, combL, statistic, grid.size, nGridSize,
    timeIGSBreakDown, takeN, verbose, timing)
```

#### Arguments

combX	The number of reads at the unique read positions	
combZ	The number of case reads at the unique read positions	
combL	The set of the labels for the unique read positions	
statistic	The type of statistic to be used. Can be 'binomial', 'rabinowitz', or 'normal'	
grid.size	The set of grid sizes for the iterative search. An automatic default can be given	
nGridSize	The number of grid sizes	
timeIGSBreakDown		
	Cumulative timing of IGS, in a broken down fashion	
takeN	The number of candidate change points to be added to a temporary set at each grid size	
verbose	If TRUE, then will print much information on each segmentation. For diagnostics only.	
timing	If TRUE, perform a timing of this algorithm, include in the output data file.	

#### Details

This algorithm is a computational speed-up tool. It computes the statistic on coarse grids, and refine to finer grids. Also, at each refinement, it computes all new smaller windows on the finer grid that would not have been captured by the coarse grid. Hence it has a New Scan step and a Refine Scan step, both implemented in C for speed. The three types of statistics are by the use of exact binomial likelihood ('binomial'), score statistic ('rabinowitz') or using normal approximation to the binomial ('normal').

#### Value

cptsRet The current set of change points called after the IGS scan of the current region timeIGSBreakDown

A break-down of the time used at the stages of the IGS

#### Author(s)

Jeremy J. Shen

#### See Also

ScanCBS, ScanStatNewComp, ScanStatRefineComp

ScanStatNewComp Main new window scan statistics computation

## Description

This is a wrapper function to call the C routines for the scan statistic new candidate segmentation computing from the IGS

#### Usage

```
ScanStatNewComp(combZCumSum, combXCumSum, combZPoint, combXPoint,
p, nTotal, grid.cur, max.win, statistic)
```

#### Arguments

combZCumSum	A cumulative sum of the number of case reads
combXCumSum	A cumulative sum of the number of reads
combZPoint	The number of case reads at the grid points
combXPoint	The number of reads at the grid points
р	The proportion of case reads in the current region
nTotal	The total number of reads in the current region
grid.cur	The current grid to be computed on
max.win	The maximum inter-window to be considered for new scan
statistic	The type of statistic. Can be 'binomial', 'rabinowitz' or 'normal'

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## ScanStatRefineComp

## Details

The computations are done in C for speed. The three types of statistics are by the use of exact binomial likelihood ('binomial'), score statistic ('rabinowitz') or using normal approximation to the binomial ('normal').

## Value

Returns a matrix containing the candidate change points from the new scan

#### Author(s)

Jeremy J. Shen

## See Also

ScanCBS, ScanIterateGrid

ScanStatRefineComp Main refining window scan statistics computation

#### Description

This is a wrapper function to call the C routines for the scan statistic to refine current candidate segmentations computing from the IGS

#### Usage

```
ScanStatRefineComp(combZCumSum, combXCumSum, combZPoint, combXPoint,
    p, nTotal, grid.cur, grid.LR, max.win, statistic)
```

#### Arguments

combZCumSum	A cumulative sum of the number of case reads
combXCumSum	A cumulative sum of the number of reads
combZPoint	The number of case reads at the grid points
combXPoint	The number of reads at the grid points
р	The proportion of case reads in the current region
nTotal	The total number of reads in the current region
grid.cur	The current grid to be computed on
grid.LR	The left and right limits of the existing candidate segmentations that will be refined, indexed by the current grid
max.win	The maximum inter-window to be considered for new scan
statistic	The type of statistic. Can be 'binomial', 'rabinowitz' or 'normal'.

#### Details

The computations are done in C for speed. The three types of statistics are by the use of exact binomial likelihood ('binomial'), score statistic ('rabinowitz') or using normal approximation to the binomial ('normal').

#### Value

Returns a matrix containing the refined candidate change points

#### Author(s)

Jeremy J. Shen

## See Also

ScanCBS, ScanIterateGrid

SegSeqResProcess Read and Process result of SegSeq

#### Description

Read the segmentation results of SegSeq and returns the change points called

#### Usage

```
SegSeqResProcess(filename)
```

### Arguments

filename The filename of the SegSeq output file to be processed

## Details

This function is used to read in the SegSeq results and use for performance evaluation and comparison

#### Value

Return a list the length of unique chromosomes in the result file. For each entry, the label is the chromosome label; and there is a vector of the change point locations called by SegSeq

#### Author(s)

Jeremy J. Shen

#### See Also

nhppSimulate

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