Package 'genomicInstability'

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Description This package contain functions to run genomic instability analysis (GIA) from scRNA-Seq data.
GIA estimates the association between gene expression and genomic location of the coding genes.
It uses the aREA algorithm to quantify the enrichment of sets of contiguous genes (loci-blocks) on the gene expression profiles and estimates the Genomic Instability Score (GIS) for each analyzed cell.

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Contents

genomicInstability-package 2
geneLength 3
genePosition 3
generateChromosomeGeneSet 4
genomicInstabilityScore 4
giDensityPlot 5
giLikelihood 6
inferCNV 7
plot.inferCNV 8

Index **10**

`genomicInstability-package`

genomicInstability: Genomic Instability estimation for scRNA-Seq

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Description

This package contain functions to run genomic instability analysis (GIA) from scRNA-Seq data. GIA estimates the association between gene expression and genomic location of the coding genes. It uses the aREA algorithm to quantify the enrichment of sets of contiguous genes (loci-blocks) on the gene expression profiles and estimates the Genomic Instability Score (GIS) for each analyzed cell.

Details

The basic functionality of this package can be performed by inferCNV(), to infer the enrichment of loci-blocks on gene expresion; genomicInstabilityScore(), to estimate the genomic instability for each of the cells in the scRNASeq dataset; giLikelihood(), to estimate the relative likelihood for each cell to be normal (low genomic instability) or tumor (high genomic instability); plot() and giDensityPlot() to plot the scores per loci-block and the distribution of the genomic instability score, respectively.

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See Also

[inferCNV()] for estimating loci-block enrichment, [genomicInstabilityScore()] for estimating the genomic instability of each cell in the dataset, [giLikelihood()] for estimating the relative likelihood for the cells to be normal or neoplastic, [plot.inferCNV()] and [giDensityPlot()] to plot the results.

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geneLength *Average length of human and mouse known genes*

Description

A dataset containing the average length for known mouse and human genes

Usage

```
geneLength
```

Format

Vector of integers indicating the average length in bp for each gene, indicated with EntrezIDs as name argument. To access this data use:

data(hg38) Human

data(mm10) Mouse

---

genePosition *Chromosomal coordinate of human and mouse known genes*

Description

A dataset containing the chromosomal coordinate for known human and mouse genes

Usage

```
genePosition
```

Format

data.frame with 2 columns: Chromosome and Coordinate. To access this data use:

data(hg38) Human

data(mm10) Mouse

generateChromosomeGeneSet *Topological gene sets*

Description

This function generates a list of sets of k genes encoded by neighbor loci

Usage

```
generateChromosomeGeneSet(species = c("human", "mouse"), k = 100, skip = 25)
```

Arguments

species	Character string indicating the species, either human or mouse
k	Integer indicating the number of genes per set
skip	Interger indicating the displacement of the window for selecting the k genes

Value

List of topoligically-close gene sets

Examples

```
chrom_set <- generateChromosomeGeneSet('human')
length(chrom_set)
chrom_set[seq_len(2)]
```

genomicInstabilityScore *Genomic Instability Analysis*

Description

This function computes the genomic instability for an object of class inferCNV

Usage

```
genomicInstabilityScore(cnv, likelihood = FALSE)
```

Arguments

cnv	Object of class inferCNV generated by inferCNV() function
likelihood	Logical, whether the genomic instability likelihood should be estimated

Value

Object of class inferCNV with updated slots for gis and gisnull

See Also

[inferCNV()] to infer the enrichment of loci-blocks in the gene expression data.

Examples

```
eh <- ExperimentHub::ExperimentHub()
dset <- eh[["EH5419"]]
tpm_matrix <- SummarizedExperiment::assays(dset)$TPM
set.seed(1)
tpm_matrix <- tpm_matrix[, sample(ncol(tpm_matrix), 500)]
cnv <- inferCNV(tpm_matrix)
cnv <- genomicInstabilityScore(cnv)
plot(density(cnv$gis))
```

---

giDensityPlot *Genomic instability plot*

Description

This function plot the genomic instability distribution, gaussian fits and null distribution if available

Usage

```
giDensityPlot(inferCNV, legend = c("topleft", "top", "topright", "none"), ...)
```

Arguments

inferCNV	Object of class inferCNV
legend	Character string indicating the location of the legend. none to not include it
...	Additional parameters for plot()

Value

None, a figure is created in the default output device

See Also

[giLikelihood()] to estimate the relative likelihood, [genomicInstabilityScore()] to estimate the genomic instability score for each cell in the dataset, and [inferCNV()] to infer the enrichment of loci-blocks in the gene expression data.

Examples

```
eh <- ExperimentHub::ExperimentHub()
dset <- eh[["EH5419"]]
tpm_matrix <- SummarizedExperiment::assays(dset)$TPM
set.seed(1)
tpm_matrix <- tpm_matrix[, sample(ncol(tpm_matrix), 500)]
cnv <- inferCNV(tpm_matrix)
cnv <- genomicInstabilityScore(cnv)
cnv <- giLikelihood(cnv, distros=c(3, 3), tumor=2:3)
giDensityPlot(cnv)
```

---

giLikelihood *Genomic instability likelihood*

Description

This function computes the genomic instability likelihood

Usage

```
giLikelihood(
  inferCNV,
  recompute = TRUE,
  distros = c(1, 3),
  tumor = NULL,
  normal = NULL
)
```

Arguments

inferCNV	InferCNV-class object
recompute	Logical, whether the model fits should be re-computed
distros	Vector of 2 integers indicating the minimum and maximum number of Gaussian models to fit
tumor	Optional vector of integers indicating the Gaussians considered as tumors
normal	Optional vector of integers indicating the Gaussians considered as normal. This is only useful when no null model has been provided for the analysis

Value

Updated inferCNV-class object with gi_likelihood slot

See Also

[genomicInstabilityScore()] to estimate the genomic instability score for each cell in the dataset, and [inferCNV()] to infer the enrichment of loci-blocks in the gene expression data.

Examples

```
eh <- ExperimentHub::ExperimentHub()
dset <- eh[["EH5419"]]
tpm_matrix <- SummarizedExperiment::assays(dset)$TPM
set.seed(1)
tpm_matrix <- tpm_matrix[, sample(ncol(tpm_matrix), 500)]
cnv <- inferCNV(tpm_matrix)
cnv <- genomicInstabilityScore(cnv)
cnv <- giLikelihood(cnv, distros=c(3, 3), tumor=2:3)
print(cnv$gi_fit)
plot(density(cnv$gi_likelihood, from=0, to=1))
```

`inferCNV` *Inference of CNV from expression data*

Description

This function estimates the CNV score based on expression data

Usage

```
inferCNV(
  expmat,
  nullmat = NULL,
  species = c("human", "mouse"),
  k = 100,
  skip = 25,
  min_geneset = 10,
  verbose = TRUE
)
```

Arguments

`expmat`	Matrix of gene expression profiles or signatures with genes '(entrezID) in rows and samples in columns
`nullmat`	Optional matrix with same number of rows as `expmat` to be used as null model
`species`	Character string indicating the species, either human or mouse
`k`	Integer indicating the number of genes per set
`skip`	Interger indicating the displacement of the window for selecting the k genes
`min_geneset`	Integer indicating the minimum size for the genesets
`verbose`	Logical, whether progress should be reported

Value

Object of class inferCNV, which is a list containing matrix of nes, and parameters (param), including species, window (k) and skip

Examples

```
eh <- ExperimentHub::ExperimentHub()
dset <- eh[["EH5419"]]
tpm_matrix <- SummarizedExperiment::assays(dset)$TPM
set.seed(1)
tpm_matrix <- tpm_matrix[, sample(ncol(tpm_matrix), 500)]
cnv <- inferCNV(tpm_matrix)
class(cnv)
names(cnv)
cnv$nes[1:5, 1:3]
```

`plot.inferCNV` *Plot chromosome map*

Description

This function generates a chromosomes map plot for the inferred CNVs

Usage

```
## S3 method for class 'inferCNV'
plot(x, output = NULL, threshold = 0.2, gamma = 1.5, resolution = 150, ...)
```

Arguments

`x`	Object of class inferCNV
`output`	Optional output PDF file name (with extension)
`threshold`	Likelihood threshold for identifying genomically inestable cells/samples, 0 disables this filter
`gamma`	Number indicating the gamma transformation for the colors
`resolution`	Integer indicating the ppi for the png and jpg output files
`...`	Additional parameters for plot

Value

Nothing, a plot is generated in the default output devise

See Also

[giLikelihood()] to estimate the relative likelihood, [genomicInstabilityScore()] to estimate the genomic instability score for each cell in the dataset, and [inferCNV()] to infer the enrichment of loci-blocks in the gene expression data.

Examples

```
eh <- ExperimentHub::ExperimentHub()
dset <- eh[["EH5419"]]
tpm_matrix <- SummarizedExperiment::assays(dset)$TPM
set.seed(1)
tpm_matrix <- tpm_matrix[, sample(ncol(tpm_matrix), 500)]
cnv <- inferCNV(tpm_matrix)
cnv <- genomicInstabilityScore(cnv)
cnv <- giLikelihood(cnv, distros=c(3, 3), tumor=2:3)
plot(cnv, output='test.png')
```

Index

∗ **datasets**
geneLength, 3
genePosition, 3
∗ **internal**
genomicInstability-package, 2

geneLength, 3
genePosition, 3
generateChromosomeGeneSet, 4
genomicInstability
*(*genomicInstability-package*)*, 2
genomicInstability-package, 2
genomicInstabilityScore, 4
giDensityPlot, 5
giLikelihood, 6

inferCNV, 7

plot.inferCNV, 8