Package ‘AllelicImbalance’

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AllelicImbalance-package

A package meant to provide all basic functions for high-throughput allele specific expression analysis

Description

Package AllelicImbalance has functions for importing, filtering and plotting high-throughput data to make an allele specific expression analysis. A major aim of this package is to provide functions to collect as much information as possible from regions of choice, and to be able to explore the allelic expression of that region in detail.

Details

Package: AllelicImbalance
Type: Package
Version: 1.2.0
Date: 2014-08-24
License: GPL-3

Overview - standard procedure

Start out creating a GRanges object defining the region of interest. This can also be done using getAreaFromGeneNames providing gene names as arguments. Then use BamImpGAList to import reads from that region and find potential SNPs using scanForHeterozygotes. Then retrieve the allele counts of heterozygote sites by the function getAlleleCount. With this data create an ASEset. At this point all pre-requisites for a 'basic' allele specific expression analysis is available. Two ways to go on could be to apply chisq.test or barplot on this ASEset object.

Author(s)

Author: Jesper Robert Gadin Author: Lasse Folkersen
Maintainer: Jesper Robert Gadin <j.r.gadin@gmail.com>

References

Reference to published application note (work in progress)

See Also

- code?ASEset
Description

These functions acts as wrappers to retrieve information from annotation database objects (annotationDb objects) or (transcriptDb objects)

Usage

genesFromAnnotation(
    OrgDb,
    GR,
    leftFlank = 0,
    rightFlank = 0,
    getUCSC = FALSE,
    verbose = FALSE
)

genesVector(OrgDb, GR, leftFlank = 0, rightFlank = 0, verbose = FALSE)

exonsFromAnnotation(
    TxDb,
    GR,
    leftFlank = 0,
    rightFlank = 0,
    verbose = FALSE
)

exonsVector(TxDb, GR, leftFlank = 0, rightFlank = 0, verbose = FALSE)

transcriptsFromAnnotation(
    TxDb,
    GR,
    leftFlank = 0,
    rightFlank = 0,
    verbose = FALSE
)

transcriptsVector(TxDb, GR, leftFlank = 0, rightFlank = 0, verbose = FALSE)

cdsFromAnnotation(TxDb, GR, leftFlank = 0, rightFlank = 0, verbose = FALSE)

cdsVector(TxDb, GR, leftFlank = 0, rightFlank = 0, verbose = FALSE)

annotationDataFrame(
    GR,
strand = "+",
annotationType = NULL,
OrgDb = NULL,
TxDb = NULL,
verbose = FALSE
}

Arguments

OrgDb An OrgDb object
GR A GenomicRanges object with sample area
leftFlank An integer specifying number of additional nucleotides around the SNPs for the leftFlank
rightFlank An integer specifying number of additional nucleotides around the SNPs for the rightFlank
getUCSC A logical indicating if UCSC transcript IDs should also be retrieved
verbose A logical making the functions more talkative
TxDb A transcriptDb object
strand Two options; '+', '-'
annotationType select one or more from 'gene', 'exon', 'transcript', 'cds'.

Details

These functions retrieve regional annotation from OrgDb or TxDb objects, when given GRanges objects.

Value

GRanges object with ranges over the genes in the region.
The getGenesVector function will return a character vector where each element are gene symbols separated by comma
GRanges object with ranges over the exons in the region.
The getTranscriptsFromAnnotation function will return a GRanges object with ranges over the transcripts in the region.
The getCDSFromAnnotation function will return a GRanges object with ranges over the CDSFs in the region.
The getExonsVector function will return a character vector where each element are exons separated by comma
The getTranscriptsVector function will return a character vector where each element are transcripts separated by comma
The getCDSVector function will return a character vector where each element are CDSs separated by comma
The getAnnotationDataFrame function will return a data.frame with annotations. This function is used internally by i.e. the barplot-function
### Description

adds a customizable annotation functionality for AllelicImbalance barplots.

### Usage

```r
annotationBarplot(
  strand,
  snp,
  lowerLeftCorner,  
  annDfPlus,  
  annDfMinus,  
  cex = 0.7, 
  ypos = 0,
  interspace = 1
)
```

### Arguments

- **strand**: strand, "+", "-", "*" or "both"
- **snp**: integer for the described SNP
- **lowerLeftCorner**: position of the plot to add legend to (default c(0,0))
Description

Generates barplots for ASEset objects. Two levels of plotting detail are provided: a detailed barplot of read counts by allele useful for fewer samples and SNPs, and a less detailed barplot of the fraction of imbalance, useful for more samples and SNPs.

Usage

```r
barplot(height, ...)  
```

## S4 method for signature 'ASEset'

```r
barplot(  
  height,  
  type = "count",  
  sampleColour.top = NULL,  
  sampleColour.bot = NULL,  
  legend = TRUE,  
  pValue = TRUE,  
  strand = "*",  
  testValue = NULL,  
  testValue2 = NULL,  
  OrgDb = NULL,  
  TxDb = NULL,  
)```
Arguments

height          An ASEset object
...             for simpler generics when extending function
type            'count' or 'fraction'
sampleColour.top User specified colours for top fraction
sampleColour.bot User specified colours for bottom fraction
legend          Display legend
pValue           Display p-value
strand          four options, '+', '-', 'both' or '*'
testValue       if set, a matrix or vector with user p-values
testValue2   if set, a matrix or vector with user p-values
OrgDb       an OrgDb object which provides annotation
TxDb        a TxDb object which provides annotation
annotationType select one or more from 'gene','exon','transcript','cds'.
main        text to use as main label
ylim        set plot y-axis limit
yaxis       whether the y-axis is to be displayed or not
xaxis       whether the x-axis is to be displayed or not
ylab        showing labels for the tic marks
ylab.text   ylab text
xlab.text   xlab text
xlab        showing labels for the tic marks
legend.colnames
            gives colnames to the legend matrix
las.ylab    orientation of ylab text
las.xlab    orientation of xlab text
cex.main    set main label size (max 2)
cex.pValue  set pValue label size
cex.ylab    set ylab label size
cex.xlab    set xlab label size
cex.legend  set legend label size
add          boolean indicates if a new device should be started
lowerLeftCorner
            integer that is only useful when add=TRUE
size        Used internally by locationplot. Rescales each small barplot window
addHorizontalLine
            adds a horizontal line that marks the default fraction of 0.5 - 0.5
add.frame   boolean to give the new plot a frame or not
filter.pValue.fraction
            numeric between 0 and 1 that filter away pValues where the main allele has this frequency.
legend.fill.size
            size of the fill/boxes in the legend (default:NULL)
legend.interspace
            set legend space between fills and text
verbose     Makes function more talkative
top.fraction.criteria
            'maxcount', 'ref' or 'phase'
cex.annotation
            size of annotation text
ypos.annotation
            relative ypos for annotation text
annotation.interspace
            space between annotation text
Details

filter.pValue.fraction is intended to remove p-value annotation with very large difference in frequency, which could just be a sequencing mistake. This is to avoid p-values like $1e^{-235}$ or similar.

sampleColourUser specified colours, either given as named colours (‘red’, ‘blue’, etc) or as hexadecimal code. Can be either length 1 for all samples, or else of a length corresponding to the number of samples for individual colouring.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

- The ASEset class which the barplot function can be called up on.

Examples

data(ASEset)
barplot(ASEset[1])

---

**ASEset-class**  
**ASEset objects**

**Description**

Object that holds allele counts, genomic positions and map-bias for a set of SNPs

**Usage**

alleleCounts(x, strand = "\*", return.class = "list")

## S4 method for signature 'ASEset'
alleleCounts(x, strand = "\*", return.class = "list")

alleleCounts(x, ...) <- value

## S4 replacement method for signature 'ASEset'
alleleCounts(x, strand = "\*", return.class = "array", ...) <- value

mapBias(x, ...)

## S4 method for signature 'ASEset'
mapBias(x, return.class = "list")

fraction(x, ...)
## S4 method for signature 'ASEset'
fraction(
  x,
  strand = "*",
  top.fraction.criteria = "maxcount",
  verbose = FALSE,
  ...
)

arank(x, return.type = "names", return.class = "list", strand = "*", ...)

frequency(x, ...)

genotype(x, ...)

## S4 method for signature 'ASEset'
genotype(x, return.class = "matrix")

genotype(x) <- value

## S4 replacement method for signature 'ASEset'
genotype(x) <- value

countsPerSnp(x, ...)

## S4 method for signature 'ASEset'
countsPerSnp(x, return.class = "matrix", return.type = "mean", strand = "*")
countsPerSample(x, ...)

## S4 method for signature 'ASEset'
countsPerSample(x, return.class = "matrix", return.type = "mean", strand = "*")

phase(x, ...)

## S4 method for signature 'ASEset'
phase(x, return.class = "matrix")

phase(x) <- value

## S4 replacement method for signature 'ASEset'
phase(x) <- value

mapBias(x) <- value

## S4 replacement method for signature 'ASEset'
mapBias(x) <- value
ASExet-class

refExist(x)
## S4 method for signature 'ASExet'
refExist(x)

ref(x)
## S4 method for signature 'ASExet'
ref(x)

ref(x) <- value
## S4 replacement method for signature 'ASExet,ANY'
ref(x) <- value

altExist(x)
## S4 method for signature 'ASExet'
altExist(x)

alt(x)
## S4 method for signature 'ASExet'
alt(x)

alt(x) <- value
## S4 replacement method for signature 'ASExet,ANY'
alt(x) <- value

aquals(x, ...)
## S4 method for signature 'ASExet'
aquals(x)

aquals(x) <- value
## S4 replacement method for signature 'ASExet'
aquals(x) <- value

maternalAllele(x, ...)
## S4 method for signature 'ASExet'
maternalAllele(x)

paternalAllele(x, ...)

## S4 method for signature 'ASEset'
paternalAllele(x)

### Arguments

- **x**: ASEset object
- **strand**: which strand of '+' or '-'
- **return.class**: return 'list' or 'array'
- **...**: additional arguments
- **value**: replacement variable
- **top.fraction.criteria**: 'maxcount', 'ref' or 'phase'
- **verbose**: makes function more talkative
- **return.type**: return 'names', rank or 'counts'

### Details

An ASEset object differs from a regular RangedSummarizedExperiment object in that the assays contains an array instead of matrix. This array has ranges on the rows, sampleNames on the columns and variants in the third dimension.

It is possible to use the commands barplot and locationplot on an ASEset object see more details in `barplot` and `locationplot`.

Three different alleleCount options are available. The simplest one is the * option, and is for experiments where the strand information is not known e.g. non-stranded data. The unknown strand could also be for strand specific data when the aligner could not find any strand associated with the read, but this should normally not happen, and if it does probably having an extremely low mapping quality. Then there are an option too add plus and minus stranded data. When using this, it is essential to make sure that the RNA-seq experiment under analysis has in fact been created so that correct strand information was obtained. The most functions will by default have their strand argument set to '*'.

The phase information is stored by the convention of 'maternal chromosome|paternal chromosome', with 0 as reference allele and 1 as alternative allele. '|' when the phase is known and '/' when the phase is unknown. Internally the information will be stored as an three dimensional array, dim 1 for SNPs, dim 2 for Samples and dim 3 which is fixed and stores maternal chromosome, paternal chromosome and phased (1 equals TRUE).

### Value

An object of class ASEset containing location information and allele counts for a number of SNPs measured in a number of samples on various strand, as well as mapBias information. All data is stored in a manner similar to the SummarizedExperiment class.

### Table

```r
table(...)
```

### Arguments:
... An ASEset object that contains the variants of interest

The generics for table does not easily allow more than one argument so in respect to the different strand options, table will return a SimpleList with length 3, one element for each strand.

**Frequency**

frequency(x, return.class = "list", strand = "*", threshold.count.sample = 15)

Arguments:
- x An ASEset object that contains the variants of interest
- threshold.count.samplesif sample has fewer counts the function return NA.

**Constructor**

ASEsetFromCountList(rowRanges, countListNonStranded = NULL, countListPlus = NULL, countListMinus = NULL, countListUnknown = NULL, colData = NULL, mapBiasExpMean = array(), verbose=FALSE, ...)

Arguments:
- rowRanges A GenomicRanges object that contains the variants of interest
- countListNonStranded A list where each entry is a matrix with allele counts as columns and sample counts as rows
- countListPlus A list where each entry is a matrix with allele counts as columns and sample counts as rows
- countListMinus A list where each entry is a matrix with allele counts as columns and sample counts as rows
- countListUnknown A list where each entry is a matrix with allele counts as columns and sample counts as rows
- colData A DataFrame object containing sample specific data
- mapBiasExpMean A 3D array describing mapping bias. The SNPs are in the 1st dimension, samples in the 2nd dimension and variants in the 3rd dimension.
- verbose Makes function more talkative
- ... arguments passed on to SummarizedExperiment constructor

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
#make example countList
set.seed(42)
countListPlus <- list()
snps <- c('snp1', 'snp2', 'snp3', 'snp4', 'snp5')
for(snp in snps){
  count<-matrix(rep(0,16),ncol=4,dimnames=list(000001
```
c('sample1','sample2','sample3','sample4'),c('A','T','G','C'))
    # insert random counts in two of the alleles
    for(allele in sample(c('A','T','G','C'),2)) {
        count[,allele] <- as.integer(rnorm(4, mean = 50, sd = 10))
    }
    countListPlus[[snp]] <- count
}

# make example rowRanges
rowRanges <- GRanges(
    seqnames = Rle(c('chr1', 'chr2', 'chr3', 'chr4')),
    ranges = IRanges(1:5, width = 1, names = head(letters, 5)),
    snp = paste('snp', 1:5, sep = '')
)

# make example colData
colData <- DataFrame(Treatment = c('ChIP', 'Input', 'Input', 'ChIP'),
    row.names = c('ind1', 'ind2', 'ind3', 'ind4'))

# make ASEset
a <- ASEsetFromCountList(rowRanges, countListPlus = countListPlus,
    colData = colData)

# example phase matrix (simple form)
p1 <- matrix(sample(c(1, 0), replace = TRUE, size = nrow(a) * ncol(a)), nrow = nrow(a),
    ncol = a)
p2 <- matrix(sample(c(1, 0), replace = TRUE, size = nrow(a) * ncol(a)), nrow = nrow(a),
    ncol = a)
p <- matrix(paste(p1, sample(c('|', '|', '/', '')[size = nrow(a) * ncol(a)], replace = TRUE),
    p2, sep = ''), nrow = nrow(a), ncol = a)
phase(a) <- p

# generate ASEset from array
snps <- 999
samples <- 5
ar <- array(rep(unlist(lapply(1:snps,
    function(x){(sample(c(TRUE, FALSE, TRUE), sample = 4).sex)})samples), samples),
    dim = c(4, snps, samples))
ar2 <- array(sample(50:300, 4*snps*samples, replace = TRUE), dim = c(4, snps, samples))
ar2[ar] <- 0
ar2 <- aperm(ar2, c(2, 3, 1))
dimnames(ar2) <- list(paste("snp", 1:snps, sep = ""), paste("sample", 1:samples, sep = ""),
    c("A", "C", "G", "T"))
gr <- GRanges(seqnames = c("chr2"), ranges = IRanges(start = 1:dim(ar2)[1], width = 1), strand = "*")
a <- ASEsetFromArrays(gr, countsUnknown = ar2)
Description

useful genotype filters

Usage

hetFilt(x, ...)

## S4 method for signature 'ASEset'
hetFilt(x, source = "genotype", ...)

Arguments

x ASSet object
...
source ‘genotype’ or ‘alleleCounts’

Details

hetFilt returns TRUE if the samples is heterozygote, based on stored genotype information present in the phase data.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

# load example data
data(ASEset)
a <- ASEset

genotype(a) <- inferGenotypes(a)
hets <- hetFilt(a)
ASEset-gbarplot

gbarplot ASEset objects

Description

Generates gbarplots for ASEset objects. Two levels of plotting detail are provided: a detailed gbarplot of read counts by allele useful for fewer samples and SNPs, and a less detailed gbarplot of the fraction of imbalance, useful for more samples and SNPs.

Usage

gbarplot(x, type = "count", strand = "*", verbose = FALSE, ...)

Arguments

x  An ASEset object

 type 'count' or 'fraction'

 strand four options, '+', '-', 'both' or '*'

 verbose Makes function more talkative

... for simpler generics when extending function

Details

This function serves the same purpose as the normal barplot, but with trellis graphics using lattice, to be able to integrate well with Gviz track functionality.

Author(s)

Jesper R. Gadin

See Also

- The ASEset class which the gbarplot function can be called up on.
- The barplot non trellis barplot.

Examples

data(ASEset)
gbarplot(ASEset[1])
ASEst-glocationplot

ASEst-glocationplot  glocationplot ASEst objects

Description
plotting ASE effects over a specific genomic region using Gviz functionality

Usage
glocationplot(
  x,
  type = "fraction",
  strand = "*",
  BamGAL = NULL,
  GenomeAxisTrack = FALSE,
  trackNameDeAn = paste("deTrack", type),
  TxDb = NULL,
  sizes = NULL,
  add = FALSE,
  verbose = FALSE,
  ...
)

Arguments
  x               an ASEst object.
  type            'fraction' or 'count'
  strand          '+','-'','*' or 'both'. This argument determines which strand is plotted. See getAlleleCounts for more information of choice of strand.
  BamGAL          GAlignmentsList covering the same genomic region as the ASEst
  GenomeAxisTrack include an genomic axis track
  trackNameDeAn   trackname for deAnnotation track
  TxDb            a TxDb object which provides annotation
  sizes           vector with the sum 1. Describes the size of the tracks
  add             add to existing plot
  verbose         if set to TRUE it makes function more talkative
  ...             arguments passed on to barplot function

Details
The glocationplot methods visualises the distribution of ASE over a larger region on one chromosome. It takes and ASEst object as well as additional information on plot type (see gbarplot), strand type (see getAlleleCounts), Annotation tracks are created from the Gviz package. It is obviously important to make sure that the genome build used is set correctly, e.g. 'hg19'. sizes has to be of the same length as the number of tracks used.
Author(s)

Jesper R. Gadin

See Also

• The ASEset class which the glocationplot function can be called up on.

Examples

```r
data(ASEset)
gnome(ASEset) <- 'hg19'
glocationplot(ASEset,strand='+' )

# for ASEsets with fewer SNPs the 'count' type plot is useful
glocationplot(ASEset,type='count',strand='+ ')
```

---

**ASEset-gviztrack**  
**ASEset-gviztrack ASEset objects**

Description

plotting ASE effects over a specific genomic region

Usage

```r
ASEDAnnotationTrack(
  x,
  GR = rowRanges(x),
  type = "fraction",
  strand = "*",
  trackName = paste("deTrack", type),
  verbose = TRUE,
  ...
)
```

```r
## S4 method for signature 'ASEset'
ASEDAnnotationTrack(
  x,
  GR = rowRanges(x),
  type = "fraction",
  strand = "*",
  trackName = paste("deTrack", type),
  verbose = TRUE,
  ...
)
```
CoverageDataTrack(
    x,
    GR = rowRanges(x),
    BamList = NULL,
    strand = NULL,
    start = NULL,
    end = NULL,
    trackNameVec = NULL,
    meanCoverage = FALSE,
    verbose = TRUE,
    ...
)

Arguments

- **x**: an ASEset object.
- **GR**: genomic range of plotting
- **type**: 'fraction' or 'count'
- **strand**: '+','-'. This argument determines which strand is plotted.
- **trackName**: name of track (ASEDAnnotationTrack)
- **verbose**: Setting verbose=TRUE gives details of procedure during function run
- **BamList**: GAlignmnentsList object of reads from the same genomic region as the ASEset
- **start**: start position of reads to be plotted
- **end**: end position of reads to be plotted
- **trackNameVec**: names of tracks (CoverageDataTrack)
- **meanCoverage**: mean of coverage over samples (CoverageGataTrack)

Details

For information of how to use these tracks in more ways, visit the Gviz package manual.

Author(s)

Jesper R. Gadin

See Also

- The ASEset class which the functions can be called up on.
Examples

```r
data(ASEset)
x <- ASEset[,1:2]
r <- reads[1:2]
genome(x) <- 'hg19'
seqlevels(r) <- seqlevels(x)

GR <- GRanges(seqnames=seqlevels(x),
ranges=IRanges(start=min(start(x)),end=max(end(x))),
strand='+', genome=genome(x))
detrack <- ASEDAnnotationTrack(x, GR=GR, type='fraction', strand='+')
covTracks <- CoverageDataTrack(x, BamList=r, strand='+')

lst <- c(detrack, covTracks)
sizes <- c(0.5, rep(0.5/length(covTracks), length(covTracks)))

# temporarily do not run this function
# plotTracks(lst, from=min(start(x)), to=max(end(x)),
# sizes=sizes, col.line = NULL, showId = FALSE, main='mainText',
# cex.main=1, title.width=1, type='histogram')
```

---

ASEset-locationplot  locationplot ASEset objects

Description

plotting ASE effects over a specific genomic region

Usage

```r
locationplot(x, ...)
```

```r
## S4 method for signature 'ASEset'
locationplot(
  x,
  type = "fraction",
  strand = "*",
  yaxis = TRUE,
  xaxis = FALSE,
  xlab = FALSE,
  ylab = TRUE,
  xlab.text = "",
  ylab.text = "",
  legend.colnames = "",
  size = c(0.8, 1),
)```
Arguments

x

... arguments passed on to barplot function

type

strand

yaxis

xaxis

xlab

ylab

xlab.text

ylab.text

legend.colnames

size

main

pValue

cex.main

cex.ylab

cex.legend

OrgDb

TxDb

verbose

top.fraction.criteria

allow.whole.chromosome

an ASEset object.

'fraction' or 'count'

'+','-','*' or 'both'. This argument determines which strand is plotted. See getAlleleCounts for more information on strand.

whether the y-axis is to be displayed or not

whether the x-axis is to be displayed or not

showing labels for the tic marks

showing labels for the tic marks

xlab text

ylab text

gives colnames to the legend matrix

will give extra space in the margins of the inner plots

text to use as main label

Display p-value

set main label size

set ylab label size

set legend label size

an OrgDb object from which to plot a gene map. If given together with argument TxDb this will only be used to extract genesymbols.

a TxDb object from which to plot an exon map.

Setting verbose=TRUE gives details of procedure during function run

'maxcount', 'ref' or 'phase'

logical, overrides 200kb region limit, defaults to FALSE
Details

The locationplot methods visualises how fractions are distributed over a larger region of genes on one chromosome. It takes and ASEset object as well as additional information on plot type (see barplot), strand type (see getAlleleCounts), colouring, as well as annotation. The annotation is taken either from the bioconductor OrgDb-sets, the TxDb sets or both. It is obviously important to make sure that the genome build used is the same as used in aligning the RNA-seq data.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

• The ASEset class which the locationplot function can be called up on.

Examples

data(ASEset)
locationplot(ASEset)

#SNPs are plotted in the order in which they are found.
#This can be sorted according to location as follows:
locationplot(ASEset[order(start(rowRanges(ASEset))],])

#for ASEsets with fewer SNPs the 'count' type plot is
# useful for detailed visualization.
locationplot(ASEset,type='count',strand='*')

---

scanForHeterozygotes

Description

Identifies the positions of SNPs found in BamGR reads.

Usage

scanForHeterozygotes(BamList, ...)

## S4 method for signature 'GAlignmentsList'
scanForHeterozygotes(
  BamList,
  minimumReadsAtPos = 20,
  maximumMajorAlleleFrequency = 0.9,
  minimumMinorAlleleFrequency = 0.1,
ASEset-scanForHeterozygotes

```r
minimumBiAllelicFrequency = 0.9,
verbose = TRUE,
```

Arguments

- **BamList**: A GAlignmentsList object
- **minimumReadsAtPos**: argument to pass on
- **minimumMajorAlleleFrequency**: minimum number of reads required to call a SNP at a given position
- **maximumMajorAlleleFrequency**: maximum frequency allowed for the most common allele. Setting this parameter lower will minimise the SNP calls resulting from technical read errors, at the cost of missing loci with potential strong ASE
- **minimumMinorAlleleFrequency**: minimum frequency allowed for the second most common allele. Setting this parameter higher will minimise the SNP calls resulting from technical read errors, at the cost of missing loci with potential strong ASE
- **minimumBiAllelicFrequency**: minimum frequency allowed for the first and second most common allele. Setting a Lower value for this parameter will minimise the identification of loci with three or more alleles in one sample. This is useful if sequencing errors are suspected to be common.
- **verbose**: logical indicating if process information should be displayed

Details

This function scans all reads stored in a GAlignmentsList for possible heterozygote positions. The user can balance the sensitivity of the search by modifying the minimumReadsAtPos, maximumMajorAlleleFrequency and minimumBiAllelicFrequency arguments.

Value

scanForHeterozygotes returns a GRanges object with the SNPs for the BamList object that was used as input.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

- The `getAlleleCounts` which is a function that count the number of reads overlapping a site.
Examples

```r
data(reads)
s <- scanForHeterozygotes(reads, verbose=FALSE)
```

---

**ASEset.old**  
*ASEset.old object*

**Description**

Old version of an ASEset which needs to be updated

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
# load example data (Not Run)
data(ASEset.old)
```

---

**ASEset.sim**  
*ASEset.sim object*

**Description**

ASEset with simulated data with SNPs within the first 200bp of chromosome 17, which is required to have example data for the refAllele function.

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
# load example data (Not Run)
data(ASEset.sim)
```
ASEsetFromBam

ASEsetFromBam

Description

count alleles and create an ASEset direct from bam file instead of reading into R first.

Usage

ASEsetFromBam(gr, ...)

## S4 method for signature 'GRanges'
ASEsetFromBam(
  gr,
  pathToDir,
  PE = TRUE,
  flagsMinusStrand = c(83, 163),
  flagsPlusStrand = c(99, 147),
  strandUnknown = FALSE,
  ...
)

Arguments

gr  GenomicRanges of SNPs to create ASEset for
...
  passed on to ASEsetFromBam function
pathToDir  Directory of bam files with index in same directory
PE  if paired end or not (default: TRUE)
flagsMinusStrand  flags that mark reads coming from minus strand
flagsPlusStrand  flags that mark reads coming from plus strand
strandUnknown  default: FALSE

Details

counts the alleles in a bam file based on GRanges positions.

Author(s)

Jesper R. Gadin
Examples

data(GRvariants)
gr <- GRvariants

# no execution at the moment
#pathToDir <- system.file('inst/extdata/ERP000101_subset', package='AllelicImbalance')
a <- ASEsetFromBam(gr, pathToDir)

Description

Generates lattice barplots for ASEset objects. Two levels of plotting detail are provided: a detailed barplot of read counts by allele useful for fewer samples and SNPs, and a less detailed barplot of the fraction of imbalance, useful for more samples and SNPs.

Usage

barplotLatticeFraction(identifier, ...)

barplotLatticeCounts(identifier, ...)

Arguments

identifier, the single snp name to plot
... used to pass on variables

Details

filter.pValue.fraction is intended to remove p-value annotation with very large difference in frequency, which could just be a sequencing mistake. This is to avoid p-values like 1e-235 or similar.
sampleColourUser specified colours, either given as named colours (‘red’, ‘blue’, etc) or as hexadecimal code. Can be either length 1 for all samples, or else of a length corresponding to the number of samples for individual colouring.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

- The ASEset class which the barplot function can be called up on.
**binom.test**

**Examples**

```r
a <- ASEset
name <- rownames(a)[1]

barplotLatticeFraction(identifier=name, x=a, astrand="+")
barplotLatticeCounts(identifier=name, x=a, astrand="+")
```

**Description**

Performs a binomial test on an ASEset object.

**Usage**

```r
## S4 method for signature 'ASEset'
binom.test(x, n = "*")
```

**Arguments**

- `x`: ASEset object
- `n`: strand option

**Details**

The test can only be applied to one strand at the time.

**Value**

`binom.test` returns a matrix

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**See Also**

- The `chisq.test` which is another test that can be applied on an ASEset object.

**Examples**

```r
# load example data
data(ASEset)

# make a binomial testinom.test(ASEset,'*')
```
chisq.test

**Description**

Performs a chisq.test on an ASEset object.

**Usage**

```r
## S4 method for signature 'ASEset'
chisq.test(x, y = "*")
```

**Arguments**

- `x` : ASEset object
- `y` : strand option

**Details**

The test is performed on one strand in an ASEset object.

**Value**

chisq.test returns a matrix with the chisq.test P-value for each SNP and sample

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**See Also**

- The `binom.test` which is another test that can be applied on an ASEset object.

**Examples**

```r
#load example data
data(ASEset)

#make a chi-square test on default non-stranded strand
chisq.test(ASEset)
```
**Description**

From a GAlignments calculate the real corresponding position for each read based on its cigar.

**Usage**

```r
realCigarPosition.old(RleCigar, BpPos)
realCigarPositions.old(RleCigar)
realCigarPositionsList.old(RleCigarList)
```

**Arguments**

- `RleCigar`: An `Rle` containing cigar information
- `BpPos`: the absolute position on the chromosome of interest
- `RleCigarList`: An `RleList` containing cigar information

**Details**

The main intention for these functions are to be the internal functions for `scanForHeterozygotes` and `getAlleleCount`.

**Value**

- `realCigarPosition` returns the new position
- `realCigarPositions` returns a vector with the corrected positions to be subsetted from a read.
- `realCigarPositionsList` returns a list where each element i a vector with the corrected positions to be subsetted from a read.

**Author(s)**

Jesper R. Gadin

**Examples**

```r
RleCigarList <- cigarToRleList('3M4I93M')
BpPos <- 5

newPos <- realCigarPosition.old(RleCigar=RleCigarList[[1]], BpPos)
newPositions <- realCigarPositions.old(RleCigar=RleCigarList[[1]])
newPositionsList <- realCigarPositionsList.old(RleCigarList=RleCigarList)
```
countAllelesFromBam

alleleCounts from bam file

Description

count alleles before creating ASEs.

Usage

countAllelesFromBam(gr, ...)

## S4 method for signature 'GRanges'
countAllelesFromBam(
gr,
pathToDir,
flag = NULL,
scanBamFlag = NULL,
return.class = "array",
verbose = TRUE,
...)

Arguments

gr  GRanges that contains SNPs of interest
...
arguments to pass on
pathToDir  path to directory of bam files
flag  specify one flag to use as filter, default is no filtering. allowed flags are 99, 147, 83 and 163
scanBamFlag  set a custom flag to use as filter
return.class  type of class for the returned object
verbose  makes function more talkative

Details

counts the alleles in a bam file based on GRanges positions.

Important excerpt from the details section of the internal applyPileups function: Regardless of 'param' values, the algorithm follows samtools by excluding reads flagged as unmapped, secondary, duplicate, or failing quality control.

Author(s)

Jesper R. Gadin
coverageMatrixListFromGAL

coverage matrix of GAlignmentsList

Description
Get coverage per nucleotide for reads covering a region

Usage
coverageMatrixListFromGAL(BamList, ...)

## S4 method for signature 'GAlignmentsList'
coverageMatrixListFromGAL(BamList, strand = "*", ignore.empty.bam.row = TRUE)

Arguments
- **BamList**: GAlignmentsList containing reads over the region to calculate coverage
- **...**: arguments to pass on
- **strand**: strand has to be '+' or '-'
- **ignore.empty.bam.row**: argument not in use atm

Details
a convenience function to get the coverage from a list of reads stored in GAlignmnetsList, and returns by default a list with one matrix, and information about the genomic start and stop positions.

Author(s)
Jesper R. Gadin

Examples
```r
r <- reads
seqlevels(r) <- '17'
covMatList <- coverageMatrixListFromGAL(BamList=r, strand='+')
```
**decorateWithExons**

**Description**

Internal function that can draw gene regions on pre-specified surfaces. Necessary for the genomic-location plots.

**Usage**

decorateWithExons(x, exonsInRegion, xlim, ylim, chromosome)

**Arguments**

- **x** ASESet object
- **exonsInRegion** GRanges object with generegions. Can be obtained using `getExonsFromAnnotation`. Must contain a column 'tx_name'
- **xlim** xlim values for the pre-specified surface
- **ylim** ylim values for the pre-specified surface
- **chromosome** character

**Details**

The main intention of this function is to be used when plotting several bar plots in the same window. This function add gene regions under the bars.

**Value**

decorateWithExons returns nothing, but draws genes

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**See Also**

- The `locationplot` which is uses this function internally.
- The `decorateWithGenes` which is another similar function that `locationplot` uses internally.

**Examples**

data(ASEset)
**decorateWithGenes**

---

**Description**

Internal function that can draw gene regions on pre-specified surfaces. Necessary for the genomic-location plots.

**Usage**

```r
decorateWithGenes(x, genesInRegion, xlim, ylim, chromosome)
```

**Arguments**

- `x` ASEset object
- `genesInRegion` GRanges object with gene regions. Can be obtained using `getGenesFromAnnotation`
- `xlim` xlim values for the pre-specified surface
- `ylim` ylim values for the pre-specified surface
- `chromosome` character

**Details**

The main intention of this function is to be used when plotting several bar plots in the same window. This function add gene regions under the bars.

**Value**

`decorateWithGenes` returns nothing, but draws genes

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**See Also**

- The `locationplot` which is uses this function internally.
- The `decorateWithExons` which is another similar function that `locationplot` uses internally.

**Examples**

```r
data(ASEset)
```
defaultMapBias

Generate default mapbias from genotype

Description

Create mapbias array from genotype matrix requires genotype information

Usage

defaultMapBias(x, ...)

## S4 method for signature 'ASEset'
defaultMapBias(x, return.class = "array")

Arguments

x ASEset object
...
internal arguments
return.class "array" or "ASEset"

Details

Default mapbias will be 0.5 for bi-allelic snps and 1 for homozygots. For genotypes with NA, 0.5 will be placed on all four alleles. Therefore tri-allelic can not be used atm. Genotype information has to be placed in the genotype(x) assay.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#load example data
data(ASEset.sim)

fasta <- system.file('extdata/hg19.chr17.subset.fa', package='AllelicImbalance')
refAllele(ASEset.sim,fasta=fasta)
a <- refAllele(ASEset.sim,fasta=fasta)
**defaultPhase**

**Description**

used to populate the phase slot in an ASEset object

**Usage**

`defaultPhase(i, ...)`

```r
## S4 method for signature 'numeric'
defaultPhase(i, j, ...)
```

**Arguments**

- `i` number of rows
- `...` arguments to forward to internal functions
- `j` number of columns

**Details**

will set everything to 0

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
i <- 5
j <- 10
defaultPhase(i, j)
```

---

**detectAI**

**Description**

detection of AllelicImbalance
Usage

detectAI(x, ...)

## S4 method for signature 'ASEset'
detectAI(
  x,
  return.class = "DetectedAI",
  strand = "*",
  threshold.frequency = 0,
  threshold.count.sample = 1,
  threshold.delta.frequency = 0,
  threshold.pvalue = 0.05,
  inferGenotype = FALSE,
  random.ref = FALSE,
  function.test = "binom.test",
  verbose = TRUE,
  gc = FALSE,
  biasMatrix = FALSE
)

Arguments

x  ASEset
...
internal arguments
return.class  class to return (atm only class 'logical')
strand  strand to infer from
threshold.frequency  least fraction to classify (see details)
threshold.count.sample  least amount of counts to try to infer allele
threshold.delta.frequency  minimum of frequency difference from 0.5 (or mapbias adjusted value)
threshold.pvalue  pvalue over this number will be filtered out
inferGenotype  infer genotypes based on count data in ASEset object
random.ref  set the reference as random if you dont know. Affects interpretation of results.
function.test  At the moment the only available option is 'binomial.test'
verbose  makes function more talkative
gc  use garbage collection when possible to save space
biasMatrix  use biasMatrix in ASEset, or use default expected frequency of 0.5 for all sites

Details

threshold.frequency is the least fraction needed to classify as bi tri or quad allelic SNPs. If 'all' then all of bi tri and quad allelic SNPs will use the same threshold. Everything under the threshold
will be regarded as noise. 'all' will return a matrix with snps as rows and uni bi tri and quad will be columns. For this function Anything that will return TRUE for tri-allelic will also return TRUE for uni and bi-allelic for the same SNP an Sample.

return.type 'ref' return only AI when reference allele is more expressed. 'alt' return only AI when alternative allele is more expressed or 'all' for both 'ref' and 'alt' alleles. Reference allele is the one present in the reference genome on the forward strand.

threshold.delta.frequency and function.test will use the value in mapBias(x) as expected value.

function.test will use the two most expressed alleles for testing. Make therefore sure there are no tri-allelic SNPs or somatic mutations among the SNPs in the ASEset.

inferGenotype(), set TRUE it should be used with as much samples as possible. If you split up the samples and run detectAI() on each sample separately, please make sure you have inferred the genotypes in before hand, alternatively used the genotypes detected by another variant Caller or chip-genotypes. Use ONLY biallelic genotypes.

Author(s)

Jesper R. Gadin

Examples

#load example data
data(ASEset)
a <- ASEset

dai <- detectAI(a)

---

DetectedAI-class

DetectedAI class

Description

Object that holds results from AI detection.

Usage

referenceFrequency(x, ...)

## S4 method for signature 'DetectedAI'
referenceFrequency(x, return.class = "array")

thresholdFrequency(x, ...)

## S4 method for signature 'DetectedAI'
thresholdFrequency(x, return.class = "array")
thresholdCountSample(x, ...)  
## S4 method for signature 'DetectedAI'
thresholdCountSample(x, return.class = "array")

thresholdDeltaFrequency(x, ...)  
## S4 method for signature 'DetectedAI'
thresholdDeltaFrequency(x, return.class = "array")

thresholdPvalue(x, ...)  
## S4 method for signature 'DetectedAI'
thresholdPvalue(x, return.class = "array")

Arguments

x: ASEset object or list of ASEsets
...
return.class: type of class returned eg. "list or "array".

Details

The DetectedAI-class contains

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

data(ASEset)
a <- ASEset
dai <- detectAI(a)

#summary(gba)
#write.tables(dai)

Description

plot functions for the DetectedAI-class
Usage

```r
frequency_vs_threshold_variable_plot(x, ...)

## S4 method for signature 'DetectedAI'
frequency_vs_threshold_variable_plot(
  x,
  var = "threshold.count.sample",
  hetOverlay = TRUE,
  smoothscatter = FALSE
)

detectedAI_vs_threshold_variable_plot(x, ...)

## S4 method for signature 'DetectedAI'
detectedAI_vs_threshold_variable_plot(
  x,
  var = "threshold.count.sample",
  summaryOverSamples = "sum",
  hetOverlay = TRUE,
  smoothscatter = FALSE
)

reference_frequency_density_vs_threshold_variable_plot(x, ...)

## S4 method for signature 'DetectedAI'
reference_frequency_density_vs_threshold_variable_plot(
  x,
  var = "threshold.count.sample"
)

detectedAI_vs_threshold_variable_multigraph_plot(x, ...)

## S4 method for signature 'DetectedAI'
detectedAI_vs_threshold_variable_multigraph_plot(x, ncol = 2, ...)

frequency_vs_threshold_variable_multigraph_plot(x, ...)

## S4 method for signature 'DetectedAI'
frequency_vs_threshold_variable_multigraph_plot(x, ncol = 2, ...)

reference_frequency_density_vs_threshold_variable_multigraph_plot(x, ...)

## S4 method for signature 'DetectedAI'
reference_frequency_density_vs_threshold_variable_multigraph_plot(
  x,
  ncol = 2,
  ...
)
```
Arguments

x  detectedAI object
...
var  string, see details for available options
hetOverlay  logical, if TRUE show nr of het SNPs used to calculate the reference allele frequency mean
smoothscatter  boolean, smoothscatter over the means
summaryOverSamples  'mean' or 'sum'
ncol  nr of columns for multiplots

Details

plot helper functions. The documentation will be improved before next release.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#some example code here
#generate example
data(ASEset)
a <- ASEset
dai <- detectAI(a,
  threshold.count.sample=1:50,
  threshold.frequency=seq(0,0.5,by=0.01),
  threshold.delta.frequency=seq(0,0.5,by=0.01),
  threshold.pvalue=rev(seq(0.001,0.05, by=0.005))
)

frequency_vs_threshold_variable_plot(dai)
detectAI_vs_threshold_variable_plot(dai)
detectAI_vs_threshold_variable_multigraph_plot(dai)
frequency_vs_threshold_variable_multigraph_plot(dai)
Usage

frequency_vs_threshold_variable_summary(x, ...)

## S4 method for signature 'DetectedAI'
frequency_vs_threshold_variable_summary(
  x,
  var = "threshold.count.sample",
  return.class = "matrix",
  ...
)

detectedAI_vs_threshold_variable_summary(x, ...)

## S4 method for signature 'DetectedAI'
detectedAI_vs_threshold_variable_summary(x, var = "threshold.count.sample")

usedSNPs_vs_threshold_variable_summary(x, ...)

## S4 method for signature 'DetectedAI'
usedSNPs_vs_threshold_variable_summary(x, var = "threshold.count.sample")

Arguments

x           detectedAI object
...          pass on variables internally
var          string, see details for available options
return.class 'matrix' or 'array'

Details

Summary helper functions. The documentation will be improved before next release.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#some example code here
#generate example
data(ASEset)
a <- ASEset
dai <- detectAI(a,
  threshold.count.sample=1:50,
  threshold.frequency=seq(0,0.5,by=0.01),
  threshold.delta.frequency=seq(0,0.5,by=0.01),
  threshold.pvalue=rev(seq(0.001,0.05, by=0.005))
)
fractionPlotDf

Description
Summarizes information to ease creating plots

Usage
fractionPlotDf(x, snp, strand = "*", top.fraction.criteria = "maxcount", ...)

## S4 method for signature 'ASEset'
fractionPlotDf(x, snp, strand = "*", top.fraction.criteria = "maxcount", ...)

Arguments
- **x**: ASEset
- **snp**: rownames identifier for ASEset or row number
- **strand**: '+', '-' or '*'
- **top.fraction.criteria**: 'maxcount', 'ref' or 'phase'
- **...**: arguments to forward to internal functions

Details
Main purpose is to reduce the amount of overall code and ease maintenance.

Top fraction criteria can take three options, maxcount, ref and phase. The top allele will be every second row in the data frame, with start from row 2. The maxcount argument will put the allele with most reads on top of the bivariate fraction. Similarly the ref argument will put always the reference allele on top. The phase arguments puts the maternal phase always on top. The top.fraction.criteria for the ref or phase arguments requires that both ref and alt is set in mcols(ASEset).

Author(s)
Jesper R. Gadin, Lasse Folkesen

Examples
#test on example ASEset
data(ASEset)
a <- ASEset
df <- fractionPlotDf(a, 1, strand="+")
gba

global analysis wrapper

Description
A wrapper to make a global analysis based on paths for BAM, VCF and GFF files

Usage
gba(pathBam, ...)

## S4 method for signature 'character'
gba(pathBam, pathVcf, pathGFF = NULL, verbose)

Arguments
- pathBam: path to bam file
- ...: arguments to pass on
- pathVcf: path to vcf file
- pathGFF: path to gff file
- verbose: makes function more talkative

Author(s)
Jesper R. Gadin

Examples
#empty as function doesn't exist

---

genomatrix

genomatrix object

Description
genomatrix is an example of a matrix with genotypes

Author(s)
Jesper R. Gadin, Lasse Folkersen

Examples
#load example data (Not Run)
#data(genomatrix)
Description

used to convert the genomatrix from the visually friendly matrix to phase array.

Usage

genotype2phase(x, ...)

## S4 method for signature 'matrix'
genotype2phase(
x, ref = NULL, return.class = "array", levels = c("A", "C", "G", "T"), ...
)

Arguments

x matrix see examples
...
ref reference alleles
return.class 'array' or 'list'
levels vector of expected alleles

Details

To not introduce redundant information in the ASEset object, the genotype matrix is translated to a phase matrix, containing the same information. Does not allow tri-allelic or multi-allelic SNPs, and if present the multi-allelic SNPs will lose the least occurring genotype.

This function can handle indels, but if the reference allele is not provided, the rank matrix which is temporary created might use lots of memory, depending on the amount of indels among the genotypes. As conclusion, it is preferable to send in reference genome when converting to phase.

levels information is only important if the reference allele has to be guessed, and so if reference information is provided, the levels argument can be ignored.

Author(s)

Jesper R. Gadin, Lasse Folkersen
getAlleleCounts

Examples

# load example data
data(genomatrix)
data(ASEset)
p <- genotype2phase(genomatrix, ref(ASEset))

getAlleleCounts snp count data

Description

Given the positions of known SNPs, this function returns allele counts from a BamGRL object

Usage

getAlleleCounts(BamList, ...)

## S4 method for signature 'GAlignmentsList'
getAlleleCounts(
  BamList,
  GRvariants,
  strand = "*",
  return.class = "list",
  verbose = TRUE,
  ...
)

Arguments

BamList A GAlignmentsList object or GRangesList object containing data imported from a bam file
...
parameters to pass on
GRvariants A GRanges object that contains positions of SNPs to retrieve
strand A length 1 character with value '+' , '-' , or '*' . This argument determines if getAlleleCounts will retrieve counts from all reads, or only from reads marked as '+' , '-' or '*' (unknown), respectively.
return.class 'list' or 'array'
verbose Setting verbose=TRUE makes function more talkative
Details

This function is used to retrieve the allele counts from specified positions in a set of RNA-seq reads. The BamList argument will typically have been created using the impBamGAL function on bam-files. The GRvariants is either a GRanges with user-specified locations or else it is generated through scanning the same bam-files as in BamList for heterozygote locations (e.g. using scanForHeterozygotes). The GRvariants will currently only accept locations having width=1, corresponding to bi-allelic SNPs. In the strand argument, specifying '*' is the same as retrieving the sum count of '+' and '-' reads (and unknown strand reads in case these are found in the bam file). '*' is the default behaviour and can be used when the RNA-seq experiments strand information is not available.

Value

g AlleleCounts returns a list of several data.frame objects, each storing the count data for one SNP.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

- The scanForHeterozygotes which is a function to find possible heterozygote sites in a GenomicAlignments object

Examples

```r
#load example data
data(reads)
data(GRvariants)

#get counts at the three positions specified in GRvariants
alleleCount <- getAlleleCounts(BamList=reads, GRvariants, strand='*')

#if the reads had contained stranded data, these two calls would have given the correct input objects for getAlleleCounts
alleleCountPlus <- getAlleleCounts(BamList=reads, GRvariants, strand='+')
alleleCountMinus <- getAlleleCounts(BamList=reads, GRvariants, strand='-')
```
Description

Given the positions of known SNPs, this function returns allele quality from a BamGRL object.

Usage

getAlleleQuality(BamList, ...)

## S4 method for signature 'GAlignmentsList'
getAlleleQuality(
  BamList,
  GRvariants,
  fastq.format = "illumina.1.8",
  return.class = "array",
  verbose = TRUE,
  ...
)

Arguments

- **BamList**: A `GAlignmentsList` object or `GRangesList` object containing data imported from a bam file.
- **...**: parameters to pass on
- **GRvariants**: A `GRanges` object that contains positions of SNPs to retrieve.
- **fastq.format**: default 'illumina.1.8'
- **return.class**: 'list' or 'array'
- **verbose**: Setting `verbose=TRUE` makes function more talkative

Details

This function is used to retrieve the allele quality strings from specified positions in a set of RNA-seq reads. The `BamList` argument will typically have been created using the `impBamGAL` function on bam-files. The `GRvariants` is either a `GRanges` with user-specified locations or else it is generated through scanning the same bam-files as in `BamList` for heterozygote locations (e.g. using `scanForHeterozygotes`). The `GRvariants` will currently only accept locations having width=1, corresponding to bi-allelic SNPs. The strand type information will be kept in the returned object. If the strand is marked as unknown "*", it will be forced to the "+" strand.

Quality information is extracted from the `BamList` object, and requires the presence of `mcols(BamList)["qual"]` to contain quality sequences.

Value

getAlleleQuality returns a list of several data.frame objects, each storing the count data for one SNP.
Author(s)
   Jesper R. Gadin, Lasse Folkersen

Examples
   # load example data
data(reads)
data(GRvariants)

   # get counts at the three positions specified in GRvariants
alleleQualityArray <- getAlleleQuality(BamList=reads,GRvariants)

   # place in ASEset object
alleleCountsArray <- getAlleleCounts(BamList=reads,GRvariants,
                           strand='*', return.class="array")

   a <- ASEsetFromArrays(GRvariants, countsUnknown = alleleCountsArray)
aquals(a) <- alleleQualityArray

getAreaFromGeneNames   Get Gene Area

Description
   Given a character vector with genesymbols and an OrgDb object, this function returns a GRanges giving the coordinates of the genes.

Usage
   getAreaFromGeneNames(genesymbols, ...)

   ## S4 method for signature 'character'
getAreaFromGeneNames(  
genesymbols,  
OrgDb,  
leftFlank = 0,  
rightFlank = 0,  
na.rm = FALSE,  
verbose = TRUE
)

Arguments
   genesymbols   A character vector that contains genesymbols of genes from which we wish to retrieve the coordinates
   ...           arguments to pass on
   OrgDb         An OrgDb object containing gene annotation
getDefaultMapBiasExpMean

leftFlank       A integer specifying number of additional nucleotides before the genes
rightFlank      A integer specifying number of additional nucleotides after the genes
na.rm           A boolean removing genes that returned NA from the annotation
verbose         Setting verbose=TRUE makes function more talkative

Details

This function is a convenience function that can be used to determine which genomic coordinates to specify to e.g. impBamGAL when retrieving reads.

The function cannot handle genes that do not exist in the annotation. To remove these please set the na.rm=TRUE.

Value

g.AreaFromGeneNames returns a GRanges object with genomic coordinates around the specified genes

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#load example data
data(ASEset)

#get counts at the three positions specified in GRvariants
library(org.Hs.eg.db )
searchArea<-getAreaFromGeneNames(c('PAX8','TLR7'), org.Hs.eg.db)

getDefaultMapBiasExpMean

Map Bias

Description

an allele frequency array

Usage

ggetDefaultMapBiasExpMean(alleleCountList, ...)

ggetDefaultMapBiasExpMean3D(alleleCountList, ...)

## S4 method for signature 'list'
ggetDefaultMapBiasExpMean(alleleCountList)
## S4 method for signature 'ANY'
getDefaultMapBiasExpMean3D(alleleCountList)

### Arguments

- **alleleCountList**
  - A GRangesList object containing read information
- ... parameters to pass on

### Details

This function will assume there is no bias that comes from the mapping of reads, and therefore create a matrix with expected frequency of 0.5 for each allele.

### Value

getDefaultMapBiasExpMean returns a matrix with a default expected mean of 0.5 for every element.

### Author(s)

Jesper R. Gadin, Lasse Folkersen

### Examples

```r
# load example data
data(ASEset)
# access SnpAfList
alleleCountList <- alleleCounts(ASEset)
# get default map bias exp mean
matExpMean <- getDefaultMapBiasExpMean(alleleCountList)
```

---

### Get snp ids from locations of SNP

**Description**

Given a GRanges object of SNPs and a SNPlocs annotation, this function attempts to replace the names of the GRanges object entries with rs-IDs.

**Usage**

```
getSnpIdFromLocation(GR, ...)
```

```r
## S4 method for signature 'GRanges'
getSnpIdFromLocation(GR, SNPloc, return.vector = FALSE, verbose = TRUE)
```
Arguments

- **GR**  
  A GRanges that contains positions of SNPs to look up

- **SNPloc**  
  A SNPlocs object containing information on SNP locations (e.g. SNPlocs.Hsapiens.dbSNP144.GRCh37)

- **return.vector**  
  Setting return.vector=TRUE returns vector with rsIds

- **verbose**  
  Setting verbose=TRUE makes function more talkative

Details

This function is used to try to identify the rs-IDs of SNPs in a GRanges object.

Value

getSnpIdFromLocation returns the same GRanges object it was given with, but with updated with rs.id information.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

```r
is_32bit_windows <- .Platform$OS.type == "windows" &&
                   .Platform$r_arch == "i386"
if (!is_32bit_windows && require(SNPlocs.Hsapiens.dbSNP144.GRCh37)) {
  #load example data
data(ASEset)

  #get counts at the three positions specified in GRvariants
  updatedGRanges <- getSnpIdFromLocation(rowRanges(ASEset),
                                          SNPlocs.Hsapiens.dbSNP144.GRCh37)
}
```

GlobalAnalysis-class  

GlobalAnalysis class

Description

Object that holds results from a global AI analysis including reference bias estimations and AI detection.

Arguments

- **x**  
  ASEset object or list of ASEsets

- **TxDb**  
  A transcriptDb object

- **...**  
  pass arguments to internal functions
Details

The GlobalAnalysis-class contains summaries and "pre-configured and pre-calculated lattice plots" needed to create an AI-report.

Value

An object of class GlobalAnalysis containing all data to make report.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

```r
data(ASEset)
#a <- ASEset
#gba <- gba(a)

#report(gba)
#write.tables(gba)
#graphs(gba)
#as.list(gba)
```

---

**GRvariants**  

*GRvariants object*

Description

This data is a GRanges object that contains the ranges for three example SNPs.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

- The `reads` which is another example object

Examples

```r
#load example data
data(GRvariants)
```
histplot

**Description**

uses base graphics hist plot

**Usage**

```r
## S4 method for signature 'ASEset'
hist(x, strand = "*", type = "mean", log = 1, ...)
```

**Arguments**

- `x` ReferenceBias object or ASEset object
- `strand` ’+’, ’-’ or ’*’
- `type` ’mean’ (only one option atm)
- `log` an integer to log each value (integer 10 for log10)
- `...` arguments to forward to interal boxplots function

**Details**

The histogram will show the density over frequencies for each sample

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
##load example data

data(ASEset)
# a <- ASEset
# hist(a)
```
**implodeList.old**

**implode list of arguments into environment**

**Description**

apply on list of variables to be put in the local environment

**Usage**

```r
implodeList.old(x)
```

**Arguments**

- `x` list of variables

**Details**

help the propagation of e.g. graphical parameters

**Author(s)**

Jesper R. Gadin

**Examples**

```r
lst <- list(hungry='yes', thirsty='no')
implodeList.old(lst)
#the check ls()
ls()
```

---

**import-bam**

**Import Bam**

**Description**

Imports a specified genomic region from a bam file using a GRanges object as search area.

**Usage**

```r
impBamGAL(UserDir, ...)
```

## S4 method for signature 'character'

```r
impBamGAL(  
  UserDir,  
  searchArea,  
  files = NULL,  
)```
import-bam

XStag = FALSE,
verbose = TRUE,
...
)

Arguments

UserDir
The relative or full path of folder containing bam files.

... arguments to pass on

searchArea
A GenomicRanges object that contains the regions of interest

files
use character vector to specify one or more files to import. The default imports all bam files from the directory.

XStag
Setting XStag=TRUE stores the strand specific information in the mcols slot 'XS'

verbose
makes the function more talkative.

Details

If the sequence data is strand-specific you may want to set XStag=TRUE. The strand specific information has then to be stored in the meta columns with column name 'XS'. If the aligner did not set the XS-tag and the data is strand-specific it is still be possible to infer the strand from the bit flags after importing the reads to R. Depending on the strand-specific protocol different combinations of the flags will have to be used. For illumina fr-secondstrand, 83 and 163 are minus strand reads and 99 and 147 are plus strand reads.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#Declare searchArea
searchArea <- GRanges(seqnames=c('17'), ranges=IRanges(79478301,79478361))

#Relative or full path
pathToFiles <- system.file('extdata/ERP000101_subset', package='AllelicImbalance')

#all files in directory
reads <- impBamGAL(pathToFiles,searchArea,verbose=FALSE)

#specified files in directory
reads <- impBamGAL(pathToFiles,searchArea,
files=c("ERR009160.bam", "ERR009167.bam"),verbose=FALSE)
Description

Imports bla bal bal a specified genomic region from a bam file using a GenomicRanges object as search area.

Usage

impBamGRL.old(UserDir, searchArea, verbose = TRUE)

Arguments

UserDir The relative or full path of folder containing bam files.
searchArea A GenomicRanges object that contains the regions of interest
verbose Setting verbose=TRUE gives details of procedure during function run.

Details

These functions are right on tahea wrappers to import bam files into R and store them into either GRanges, GAlignments or GappedAlignmentpairs objects.

It is recommended to use the impBamGAL() which takes information of gaps into account. It is also possible to use the other variants as well, but then pre-filtering becomes important keeps to understand because gapped, intron-spanning reads will cause problems. This is because the GRanges objects can not handle if gaps are present and will then give a wrong result when calculating the allele (SNP) count table.

Value

impBamGRL returns a GRangesList object containing the RNA-seq reads in the region defined by the searchArea argument. impBamGAL returns a list with GAlignments objects containing the RNA-seq reads in the region defined by the searchArea argument. funImpBamGAPL returns a list with GappedAlignmentPairs object containing the RNA-seq reads in the region defined by the searchArea argument.

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#Declare searchArea
searchArea <- GRanges(seqnames=c('17'), ranges=IRanges(79478301,79478361))

#Relative or full path
pathToFiles <- system.file('extdata/ERP000101_subset', package='AllelicImbalance')
**import-bcf**  

**Import Bcf Selection**

**Description**
Imports a selection of a bcf file or files specified by a GenomicRanges object as search area.

**Usage**
```r
impBcfGRL(UserDir, ...)  
## S4 method for signature 'character'
impBcfGRL(UserDir, searchArea = NULL, verbose = TRUE, ...)
impBcfGR(UserDir, ...)
## S4 method for signature 'character'
impBcfGR(UserDir, searchArea = NULL, verbose = TRUE, ...)
```

**Arguments**
- **UserDir**  
The relative or full path of folder containing bam files.
- **...**  
parameters to pass on
- **searchArea**  
A GenomicRanges object that contains the regions of interest
- **verbose**  
Setting verbose=TRUE gives details of the procedure during function run.

**Details**
A wrapper to import bcf files into R in the form of GenomicRanges objects.

**Value**
- BcfImpGRL returns a GRangesList object. BcfImpGR returns one GRanges object of all unique entries from one or more bcf files.

**Note**
Make sure there is a complementary index file *.bcf.csi for each bcf file in UserDir. If there is not, then the functions impBcfGRL and impBcfGR will try to create them.

**Author(s)**
Jesper R. Gadin, Lasse Folkersen
See Also

- The \texttt{impBamGRL} for importing \texttt{bam} files
- The \texttt{getAlleleCounts} for how to get allele(SNP) counts
- The \texttt{scanForHeterozygotes} for how to find possible heterozygote positions

Examples

```r
# Declare searchArea
searchArea <- GRanges(seqnames=c('17'), ranges=IRanges(79478301, 79478361))

# Relative or full path
pathToFiles <- system.file('extdata/ERP000101_subset', package='AllelicImbalance')

# import
reads <- impBcfGRL(pathToFiles, searchArea, verbose=FALSE)
```

---

\texttt{inferAlleles} \hspace{1cm} \textit{inference of SNPs of ASEset}

\textbf{Description}

\textit{inference of SNPs}

\textbf{Usage}

```
inferAlleles(
  x,
  strand = '*',
  return.type = "bi",
  threshold.frequency = 0,
  threshold.count.sample = 1,
  inferOver = "eachSample",
  allow.NA = FALSE
)
```

\textbf{Arguments}

\begin{itemize}
  \item \texttt{x} \hspace{1cm} ASEset
  \item \texttt{strand} \hspace{1cm} strand to infer from
  \item \texttt{return.type} \hspace{1cm} 'uni' 'bi' 'tri' 'quad' 'all'
  \item \texttt{threshold.frequency} \hspace{1cm} least fraction to classify (see details)
  \item \texttt{threshold.count.sample} \hspace{1cm} least amount of counts to try to infer allele
  \item \texttt{inferOver} \hspace{1cm} 'eachSample' or 'allSamples'
  \item \texttt{allow.NA} \hspace{1cm} treat NA as zero when TRUE
\end{itemize}
**inference of the alternate allele based on count data**

**Arguments**

- `x`: matrix, see examples
- `return.class`: class of returned object
- `allele.source`: 'arank'
- `verbose`: make function more talkative
- `...`: arguments to forward to internal functions

**Details**

The inference essentially ranks all alleles and the most expressed allele not declared as reference will be inferred as the alternative allele. At the moment only inference of bi-allelic alternative alleles are available.

**Author(s)**

Jesper R. Gadin, Lasse Folkeisen
Examples

```r
#load data
data(ASEset)

alt <- inferAltAllele(ASEset)
```

---

**inferGenotypes**

*inference of genotypes from ASEset count data*

### Description

inference of genotypes

### Usage

```r
inferGenotypes(
  x,  # ASEset
  strand = "*",  # strand to infer from
  return.class = "matrix",  # 'matrix' or 'vector'
  return.allele.allowed = "bi",  # vector with 'bi' 'tri' or 'quad'. 'uni' Always gets returned
  threshold.frequency = 0,  # least fraction to classify (see details)
  threshold.count.sample = 1  # least amount of counts to try to infer allele
)
```

### Arguments

- **x** 
  ASEset
- **strand** 
  strand to infer from
- **return.class** 
  'matrix' or 'vector'
- **return.allele.allowed** 
  vector with 'bi' 'tri' or 'quad'. 'uni' Always gets returned
- **threshold.frequency** 
  least fraction to classify (see details)
- **threshold.count.sample** 
  least amount of counts to try to infer allele

### Details

Often necessary information to link AI to SNPs outside coding region

### Author(s)

Jesper R. Gadin
Examples

```r
data(ASEset)
g <- inferGenotypes(ASEset)
```

Description

Functions to construct ASEset objects

Usage

```r
ASEsetFromCountList(
  rowRanges,
  countListUnknown = NULL,
  countListPlus = NULL,
  countListMinus = NULL,
  colData = NULL,
  mapBiasExpMean = NULL,
  phase = NULL,
  aquals = NULL,
  verbose = FALSE,
  ...
)
```

```r
ASEsetFromArrays(
  rowRanges,
  countsUnknown = NULL,
  countsPlus = NULL,
  countsMinus = NULL,
  colData = NULL,
  mapBiasExpMean = NULL,
  phase = NULL,
  genotype = NULL,
  aquals = NULL,
  verbose = FALSE,
  ...
)
```

Arguments

- `rowRanges`: A GenomicRanges object that contains the variants of interest
- `countListUnknown`: A list where each entry is a matrix with allele counts as columns and sample counts as rows
countListPlus  A list where each entry is a matrix with allele counts as columns and sample counts as rows
countListMinus A list where each entry is a matrix with allele counts as columns and sample counts as rows
colData       A DataFrame object containing sample specific data
mapBiasExpMean A 3D array where the SNPs are in the 1st dimension, samples in the 2nd dimension and variants in the 3rd dimension.
phase         A matrix or an array containing phase information.
aquals        A 4-D array containing the count information, see details
verbose       Makes function more talkative
...           arguments passed on to SummarizedExperiment constructor
countsUnknown An array containing the count information
countsPlus    An array containing the count information
countsMinus   An array containing the count information
genotype      matrix

Details

The resulting ASEset object is based on the RangedSummarizedExperiment class, and will therefore inherit the same accessors and ranges operations.

If both countListPlus and countListMinus are given they will be used to calculate countListUnknown, which is the sum of the plus and minus strands.

countListPlus, countListMinus and countListUnknown are i.e. the outputs from the getAlleleCounts function.

aquals is new for the devel branch and will be changed slightly before the release to include better granularity.

Value

ASEsetFromCountList returns an ASEset object.

Note

ASEsetFromCountList requires the same input data as a RangedSummarizedExperiment, but with minimum one assay for the allele counts.

Author(s)

Jesper R. Gadin, Lasse Folkersen
Examples

```r
#make example alleleCountListPlus
set.seed(42)
countListPlus <- list()
snps <- c('snp1','snp2','snp3','snp4','snp5')
for(snp in snps){
count<-matrix(rep(0,16),ncol=4,dimnames=list(
c('sample1','sample2','sample3','sample4'),
c('A','T','G','C'))) #insert random counts in two of the alleles
for(allele in sample(c('A','T','G','C'),2)){
count[,allele]<-as.integer(rnorm(4,mean=50,sd=10))
}
countListPlus[[snp]] <- count}

#make example alleleCountListMinus
countListMinus <- list()
snps <- c('snp1','snp2','snp3','snp4','snp5')
for(snp in snps){
count<-matrix(rep(0,16),ncol=4,dimnames=list(
c('sample1','sample2','sample3','sample4'),
c('A','T','G','C'))) #insert random counts in two of the alleles
for(allele in sample(c('A','T','G','C'),2)){
count[,allele]<-as.integer(rnorm(4,mean=50,sd=10))
}
countListMinus[[snp]] <- count}

#make example rowRanges
rowRanges <- GRanges(
seqnames = Rle(c('chr1', 'chr2', 'chr3', 'chr1')),
ranges = IRanges(1:5, width = 1, names = head(letters,5)),
snp = paste('snp',1:5,sep=''))

#make example colData
colData <- DataFrame(Treatment=c('ChIP', 'Input','Input','ChIP'),
row.names=c('ind1','ind2','ind3','ind4'))

#make ASEset
a <- ASEsetFromCountList(rowRanges, countListPlus=countListPlus,
countListMinus=countListMinus, colData=colData)
```
Description

Functions to construct DetectedAI objects

Usage

DetectedAIFromArray(
  x = "ASEset",
  strand = "x",
  reference.frequency = NULL,
  threshold.frequency = NULL,
  threshold.count.sample = NULL,
  threshold.delta.frequency = NULL,
  threshold.pvalue = NULL,
  threshold.frequency.names = NULL,
  threshold.count.sample.names = NULL,
  threshold.delta.frequency.names = NULL,
  threshold.pvalue.names = NULL,
  ...
)

Arguments

x ASEset
strand set strand to detectAI over "+","-","x"
reference.frequency
destribution frequencies of reference alleles based allele counts
threshold.frequency
logical array for frequency thresholds
threshold.count.sample
logical array for per sample allele count thresholds
threshold.delta.frequency
logical array for delta frequency thresholds.
threshold.pvalue
logical array for pvalue thresholds (max 1, min 0)
threshold.frequency.names
character vector
threshold.count.sample.names
character vector
threshold.delta.frequency.names
character vector
threshold.pvalue.names
character vector
... internal arguments

Details

produces a class container for reference bias calculations
Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

```r
data(ASEset)
a <- ASEset
dai <- detectAI(a)
```

---

**Initialize GlobalAnalysis**

*Initialize GlobalAnalysis*

**Description**

Functions to construct GlobalAnalysis objects

**Usage**

```r
GAnalysis(x = "ASEset", ...)
```

**Arguments**

- `x` : ASEset
- `...` : internal arguments

**Details**

produces a class container for a global analysis

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

```r
data(ASEset)
a <- ASEset
# gba <- gba(a)
```
initialize-RiskVariant

*Initialize RiskVariant*

**Description**

Functions to construct RiskVariant objects

**Usage**

RiskVariantFromGRangesAndPhaseArray(x, phase, ...)

**Arguments**

- **x**: GRanges object for the SNPs
- **phase**: array with phaseinfo
- **...**: internal arguments

**Details**

produces a class container for reference bias calculations

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

data(ASEset)
#p <- getPhaseFromSomewhere
#rv <- RiskVariantFromGRangesAndPhaseArray(x=GRvariants, phase=p)

---

**legendBarplot**: add legend to AllelicImbalance barplot

**Description**

adds a very customizable legend function for AllelicImbalance barplots.
Usage

legendBarplot(
  lowerLeftCorner,
  size,
  rownames,
  colnames,
  boxsize = 1,
  boxspace = 1,
  fgCol,
  bgCol,
  ylegendPos = 1,
  xlegendPos = 0.96,
  cex = 1
)

Arguments

lowerLeftCorner    position of the plot to add legend to (default c(0,0))
size               scale the plot, default is 1
rownames           rownames in legend
colnames           colnames in legend
boxsize            size of each box fill
boxspace           space inbetween the box fill
fgCol              color for allele1
bgCol              color for allele2
ylegendPos         placement of the legend within the plot for y
xlegendPos         placement of the legend within the plot for x
cex                size of legend text

Details

the function is preferably called from within the AllelicImbalance barplot method.

Author(s)

Jesper R. Gadin

Examples

#code placeholders
#< create a barplot with legend >
#< add legend >
LinkVariantAlmlof-class

**Description**

Object that holds results from AI detection.

**Usage**

\[ pvalue(x, \ldots) \]

```r
## S4 method for signature 'LinkVariantAlmlof'
pvalue(x)
```

**Arguments**

- `x`: LinkVariantAlmlof object
- `\ldots`: pass arguments to internal functions

**Details**

The LinkVariantAlmlof-class contains

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
# some code
```

LinkVariantAlmlof-plot

**Description**

plot an object of type LinkVariantAlmlof

**Usage**

\[ plot(x, y, \ldots) \]

```r
## S4 method for signature 'LinkVariantAlmlof,ANY'
plot(x, y, \ldots)
```
Arguments

- **x**: LinkVariantAlmlof object
- **y**: not used
- ... pass on arguments to internal methods

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

```r
data(ASEset)
a <- ASEset
# Add phase
set.seed(1)
p1 <- matrix(sample(c(1,0), replace=TRUE, size=nrow(a)*ncol(a)), nrow=nrow(a), ncol(a))
p2 <- matrix(sample(c(1,0), replace=TRUE, size=nrow(a)*ncol(a)), nrow=nrow(a), ncol(a))
p <- matrix(paste(p1, sample(c("|","|","/"), size=nrow(a)*ncol(a), replace=TRUE), p2, sep=""), nrow=nrow(a), ncol(a))

phase(a) <- p

# add alternative allele information
mcols(a)[["alt"]]<- inferAltAllele(a)

# init risk variants
p.ar <- phaseMatrix2Array(p)
rv <- RiskVariantFromGRangesAndPhaseArray(x=GRvariants, phase=p.ar)

# colnames has to be same and same order in ASEset and RiskVariant
colnames(a) <- colnames(rv)

# in this example each and every snp in the ASEset defines a region
r1 <- granges(a)

# in this example two overlapping subsets of snps in the ASEset defines the region
r2 <- split(granges(a)[c(1,2,2,3)], c(1,1,2,2))

# link variant almlof (lva)
lv1 <- lva(a, rv, r1)
lv2 <- lva(a, rv, r2)
plot(lv2[[1]])
```

Description

make an almlof regression for arrays
Usage

`lva(x, ...)

# S4 method for signature 'ASEset'
lva(
  x,
  rv,
  region,
  settings = list(),
  return.class = "LinkVariantAlmlof",
  type = "lm",
  verbose = FALSE,
  covariates = matrix(),
  ...
)

Arguments

- `x` ASESet object with phase and 'ref'/'alt' allele information
- `...` arguments to forward to internal functions
- `rv` RiskVariant object with phase and 'ref'/'alt' allele information
- `region` RiskVariant object with phase and alternative allele information
- `settings` RiskVariant object with phase and alternative allele information
- `return.class` 'LinkVariantAlmlof' (more options in future)
- `type` "lm" or "nlme", "nlme" needs subject information
- `verbose` logical, if set TRUE, then function will be more talkative
- `covariates` add data.frame with covariates (only integers and numeric)

Details

internal method that takes one array with results from regionSummary and one matrix with group information for each risk SNP (based on phase)

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

data(ASEset)
a <- ASEset
# Add phase
set.seed(1)
p1 <- matrix(sample(c(1,0), replace=TRUE, size=nrow(a)*ncol(a)), nrow=nrow(a), ncol(a))
p2 <- matrix(sample(c(1,0), replace=TRUE, size=nrow(a)*ncol(a)), nrow=nrow(a), ncol(a))
p <- matrix(paste(p1, sample(c(1,1,0,0), size=nrow(a)*ncol(a), replace=TRUE), p2, sep=""),
nrow=nrow(a), ncol(a))
phase(a) <- p

# add alternative allele information
mcols(a)[["alt"]]<- inferAltAllele(a)

# init risk variants
p.ar <- phaseMatrix2Array(p)
rv <- RiskVariantFromGRangesAndPhaseArray(x=GRvariants, phase=p.ar)

# colnames has to be same and same order in ASEset and RiskVariant
colnames(a) <- colnames(rv)

# in this example each and every snp in the ASEset defines a region
r1 <- granges(a)

# use GRangesList to merge and use regions defined by each element of the
# GRangesList
r1b <- GRangesList(r1)
r1c <- GRangesList(r1, r1)

# in this example two overlapping subsets of snps in the ASEset defines the region
r2 <- split(granges(a)[c(1, 2, 2, 3), c(1, 1, 2, 2)])

# link variant almlof (lva)
lva(a, rv, r1)
lva(a, rv, r1b)
lva(a, rv, r1c)
lva(a, rv, r2)

# Use covariates (integers or numeric)
cov <- data.frame(age=sample(20:70, ncol(a)), sex=rep(c(1, 2), each=ncol(a)/2),
                  row.names=colnames(a))
lva(a, rv, r1, covariates=cov)
lva(a, rv, r1b, covariates=cov)
lva(a, rv, r1c, covariates=cov)
lva(a, rv, r2, covariates=cov)

# link variant almlof (lva), using nlme
a2 <- a
ac <- assays(a2)[["countsPlus"]]
jit <- sample(c(seq(-0.10, 0, length=5), seq(0, 0.10, length=5)), size=length(ac), replace=TRUE)
assays(a2, withDimnames=FALSE)[["countsPlus"]]<- round(ac * (1+jit), 0)
ab <- cbind(a, a2)
colData(ab)[["subject.group"]]<- c(1:ncol(a), 1:ncol(a))
rv2 <- rv[, c(1:ncol(a), 1:ncol(a))]
colnames(ab) <- colnames(rv2)
lva(ab, rv2, r1, type="nlme")
lva(ab, rv2, r1b, type="nlme")
lva(ab, rv2, r1c, type="nlme")
lva(ab, rv2, r2, type="nlme")
Description

make an almlof regression for arrays (internal function)

Usage

lva.internal(x, ...)

## S4 method for signature 'array'
lva.internal(
x, grp, 
element = 3, 
type = "lm", 
subject = NULL, 
covariates = matrix(), 
... 
)

Arguments

x regionSummary array phased for maternal allele

... arguments to forward to internal functions

grp group 1-3 (1 for 0:0, 2 for 1:0 or 0:1, and 3 for 1:1)

element which column in x contains the values to use with lm.

type which column in x contains the values to use with lm.

subject which samples belongs to the same individual

covariates add data.frame with covariates (only integers and numeric)

Details

internal method that takes one array with results from regionSummary and one matrix with group information for each risk SNP (based on phase). Input and output objects can change format slightly in future.

Author(s)

Jesper R. Gadin, Lasse Folkersen
Examples

data(ASEset)
a <- ASEset
# Add phase
set.seed(1)
p1 <- matrix(sample(c(1,0),replace=TRUE, size=nrow(a)*ncol(a)),nrow=nrow(a), ncol(a))
p2 <- matrix(sample(c(1,0),replace=TRUE, size=nrow(a)*ncol(a)),nrow=nrow(a), ncol(a))
p <- matrix(paste(p1,sample(c("|","|","/"), size=nrow(a)*ncol(a), replace=TRUE), p2, sep=""), nrow=nrow(a), ncol(a))

phase(a) <- p

#add alternative allele information
mcols(a)[["alt"]]<- inferAltAllele(a)

# in this example two overlapping subsets of snps in the ASEset defines the region
region <- split(granges(a)[c(1,2,2,3)], c(1,1,2,2))
rs <- regionSummary(a, region, return.class="array", return.meta=FALSE)

# use (change to generated riskSNP phase later)
phs <- array(c(phase(a,return.class="array")[1,,c(1, 2)],
              phase(a,return.class="array")[2,,c(1, 2)]), dim=c(20,2,2))
grp <- matrix(2, nrow=dim(phs)[1], ncol=dim(phs)[2])
grp[(phs[,,1] == 0) & (phs[,,2] == 0)] <- 1
grp[(phs[,,1] == 1) & (phs[,,2] == 1)] <- 3
#only use mean.fr at the moment, which is col 3
lva.internal(x=assays(rs)[["rs1"]],grp=grp, element=3)

makeMaskedFasta makes masked fasta reference

Description

Replaces all selected positions in a fasta file with the character N

Usage

makeMaskedFasta(fastaIn, ...)
mapBiasRef

Arguments

fastaIn character string of the path for the fasta file to be used
... arguments to pass on
fastaOut character string of the path for the masked fasta file (no extension)
posToReplace GRanges object with the genomic ranges to replace
splitOnSeqlevels write on file for each seqlevel to save memory
verbose makes function more talkative

Author(s)

Jesper R. Gadin

Examples

data(ASEset.sim)
gr <- rowRanges(ASEset.sim)
fastaIn <- system.file('extdata/hg19.chr17.subset.fa', package='AllelicImbalance')
makeMaskedFasta(fastaIn=fastaIn, fastaOut="fastaOut", posToReplace=gr)

mapBiasRef

mapBias for reference allele

Description

Create a matrix of bias for the reference allele

Usage

mapBiasRef(x, ...)

## S4 method for signature 'ASEset'
mapBiasRef(x)

Arguments

x ASEset object
... internal arguments

Details

select the expected frequency for the reference allele
**Description**

filter on `minCountFilt` snps

**Usage**

```r
minCountFilt(x, ...)  
## S4 method for signature 'ASEset'
minCountFilt(  
  x,
  strand = "*",
  threshold.counts = 1,
  sum = "all",
  replace.with = "zero",
  return.class = "ASEset"
)
```

**Arguments**

- `x`: ASEset object
- `...`: internal param
- `strand`: strand to infer from
- `threshold.counts`: cutoff for read counts (see details)
- `sum`: 'each' or 'all'
- `replace.with`: only option 'zero'
- `return.class`: 'ASEset', 'array' or 'matrix'

**Details**

Description info here
**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
# load example data
data(ASEset)
a <- ASEset

minFreqFilt(a)
```

---

**Description**

filter on minFreqFilt snps

**Usage**

```r
minFreqFilt(x, ...)
```

## S4 method for signature 'ASEset'

```r
minFreqFilt(
  x,
  strand = "*",
  threshold.frequency = 0.1,
  replace.with = "zero",
  return.class = "ASEset",
  sum = "all"
)
```

**Arguments**

- `x`: ASEset object
- `...`: internal param
- `strand`: strand to infer from
- `threshold.frequency`: least fraction to classify (see details)
- `replace.with`: only option 'zero'
- `return.class`: 'ASEset', 'array' or 'matrix'
- `sum`: 'each' or 'all'

**Details**

Description info here
multiAllelicFilt

Author(s)

Jesper R. Gadin, Lasse Folkesen

Examples

# load example data
data(ASEset)
a <- ASEset

minFreqFilt(a)

multiAllelicFilt

multi-allelic filter methods

Description

filter on multiallelic snps

Usage

multiAllelicFilt(x, ...)

## S4 method for signature 'ASEset'
multiAllelicFilt(
x,
strand = "*",
threshold.count.sample = 10,
threshold.frequency = 0.1,
filterOver = "eachSample"
)

Arguments

x ASEset object
...
internal param
strand strand to infer from
threshold.count.sample
least amount of counts to try to infer allele
threshold.frequency
least fraction to classify (see details)
filterOver 'eachSample' or 'allSamples'

Details

based on the allele counts for all four variants A, T, G and C and returns true if there is counts enough suggesting a third or more alleles. The sensitivity can be specified using 'threshold.count.sample' and 'threshold.frequency'.
Author(s)

Jesper R. Gadin, Lasse Folkeren

Examples

#load example data
data(ASEset)
a <- ASEset
multiAllelicFilt(a)

Description

Convert the phase from the internally stored phase, ref and alt information

Usage

phase2genotype(x, ...)

## S4 method for signature 'array'
phase2genotype(x, ref, alt, return.class = "matrix", ...)

Arguments

x
array see examples

... pass on additional param

ref reference allele vector

alt alternative allele vector

return.class 'matrix' or 'array'

Details

To not introduce redundant information in the ASEset object, the genotype matrix is accessed from the phase matrix, which together with ref and alt allele information contains the same information (not taken into account three-allelic or more SNPs).

The genotype matrix retrieved from an ASEset object can differ from the genotype matrix stored in the object if reference and alternative alleles were not used or has changed since the phase genotype matrix was stored. Basically, it is preferable to provide reference and alternative information when storing the genotype matrix.

If possible, it is better to not use a genotype matrix, but instead relying completely on storing a phase matrix (or array) together with reference and alternative allele information.
**phaseArray2phaseMatrix**

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
# load example data
data(ASEset)
data(genomatrix)
p <- genotype2phase(genomatrix, ref(ASEset), return.class="array")
ref <- ref(ASEset)
alt <- inferAltAllele(ASEset)

gt <- phase2genotype(p, ref, alt, return.class="matrix")
```

**Description**

used to convert the phase from the visually friendly matrix to array.

**Usage**

```r
phaseArray2phaseMatrix(x, ...)
```

```r
## S4 method for signature 'array'
phaseArray2phaseMatrix(x, ...)
```

**Arguments**

- `x` array see examples
- `...` arguments to forward to internal functions

**Details**

A more effectice way of store the phase data in the ASEset object

**Author(s)**

Jesper R. Gadin, Lasse Folkersen
Examples

```r
#load data
data(ASEset)
a <- ASEset

#example phase matrix
p1 <- matrix(sample(c(1,0),replace=TRUE, size=nrow(a)*ncol(a)),nrow=nrow(a), ncol(a))
p2 <- matrix(sample(c(1,0),replace=TRUE, size=nrow(a)*ncol(a)),nrow=nrow(a), ncol(a))
p <- matrix(paste(p1,sample(c("|","|","/"), size=nrow(a)*ncol(a), replace=TRUE), p2, sep=""),
nrow=nrow(a), ncol(a))

ar <- phaseMatrix2Array(p)

#Convert back
mat <- phaseArray2phaseMatrix(ar)
```

Description

used to convert the phase from the visually friendly matrix to array.

Usage

```r
phaseMatrix2Array(x, ...)
```

## S4 method for signature 'matrix'
phaseMatrix2Array(x, dimnames = NULL, ...)

Arguments

- `x`   matrix see examples
- `...` arguments to forward to internal functions
- `dimnames` list with dimnames

Details

A more effective way of store the phase data in the ASEset object

Author(s)

Jesper R. Gadin, Lasse Folkersen
Examples

# load data
data(ASEset)
a <- ASEset

# example phase matrix
p1 <- matrix(sample(c(1,0), replace=TRUE, size=nrow(a)*ncol(a)), nrow=nrow(a), ncol(a))
p2 <- matrix(sample(c(1,0), replace=TRUE, size=nrow(a)*ncol(a)), nrow=nrow(a), ncol(a))
p <- matrix(paste(p1, sample(c("|","|","/"), size=nrow(a)*ncol(a), replace=TRUE), p2, sep=""),
nrow=nrow(a), ncol(a))
ar <- phaseMatrix2Array(p)

randomRef

Random ref allele from genotype

Description

Create a vector of random reference alleles

Usage

randomRef(x, ...)

## S4 method for signature 'ASEset'
randomRef(x, source = "alleleCounts", ...)

Arguments

x

ASEset object

...

internal arguments

source

'alleleCounts'

Details

Randomly shuffles which of the two alleles for each genotype that is indicated as reference allele,

based on either allele count information or previous ref and alt alleles.

When the source is 'alleleCounts', the two most expressed alleles are taken as reference and alternative allele.

Author(s)

Jesper R. Gadin, Lasse Folkersen
Examples

```r
# load example data
data(ASEset.sim)
a <- ASEset.sim

ref(a) <- randomRef(a, source = 'alleleCounts')
```

reads

**reads object**

Description

This data set corresponds to the BAM-file data import illustrated in the vignette. The data set consists of a chromosome 17 region from 20 RNA-seq experiments of HapMap samples.

Author(s)

Jesper R. Gadin, Lasse Folkersen

References


See Also

- The `GRvariants` which is another example object

Examples

```r
## load example data (Not Run)
data(reads)
```

refAllele

**Reference allele**

Description

Extract the allele based on SNP location from the reference fasta file

Usage

```r
refAllele(x, fasta)
```
regionSummary

Arguments

x  ASEset object
fasta  path to fasta file, index should be located in the same folder

Details

The alleles will be placed in the rowRanges() meta column 'ref'

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

# load example data
data(ASEset.sim)

fasta <- system.file('extdata/hg19 chr17 subset fa', package='AllelicImbalance')
a <- refAllele(ASEset.sim, fasta=fasta)

regionSummary

Description

Gives a summary of AI-consistency for a transcript

Usage

regionSummary(x, ...)

## S4 method for signature 'ASEset'
regionSummary(x, region, strand = "*", return.class = "RegionSummary", ...)

Arguments

x  ASEset object
...  arguments to forward to internal functions
region  to summarize over, the object can be a GRanges, GRangesList
strand  can be "+", "-" or "*
return.class  "array" or "list".
RegionSummary-class

Description

Object that holds results from the regionSummary method
**Usage**

```r
sumnames(x, ...)
```

## S4 method for signature 'RegionSummary'

sumnames(x)

```r
basic(x, ...)
```

## S4 method for signature 'RegionSummary'

basic(x)

**Arguments**

- `x` : RegionSummary object
- `...` : pass arguments to internal functions

**Details**

The `RegionSummary` class objects contain summaries for specified regions.

**Author(s)**

Jesper R. Gadin, Lasse Folkersen

**Examples**

```r
#some code
```

---

**RiskVariant-class**  

**RiskVariant class**

**Description**

Object that holds results from AI detection.

**Usage**

```r
## S4 method for signature 'RiskVariant'
ref(x)
```

```r
## S4 replacement method for signature 'RiskVariant,ANY'
ref(x) <- value
```

```r
## S4 method for signature 'RiskVariant'
alt(x)
```
scanForHeterozygotes.old

## S4 replacement method for signature 'RiskVariant,ANY'
alt(x) <- value

## S4 method for signature 'RiskVariant'
phase(x, return.class = "matrix")

## S4 replacement method for signature 'RiskVariant'
phase(x) <- value

Arguments

x RiskVariant object or list of RiskVariants
value argument used for replacement
return.class type of class returned eg. "list" or "array".

Details

The RiskVariant-class contains

Author(s)

Jesper R. Gadin, Lasse Folkersen

Examples

#some code

Description

Identifies the positions of SNPs found in BamGR reads.

Usage

scanForHeterozygotes.old(
  BamList,
  minimumReadsAtPos = 20,
  maximumMajorAlleleFrequency = 0.9,
  minimumBiAllelicFrequency = 0.9,
  maxReads = 15000,
  verbose = TRUE
)
scanForHeterozygotes.old

Arguments

BamList A GAlignmentsList object
minimumReadsAtPos minimum number of reads required to call a SNP at a given position
maximumMajorAlleleFrequency maximum frequency allowed for the most common allele. Setting this parameter lower will minimise the SNP calls resulting from technical read errors, at the cost of missing loci with potential strong ASE
minimumBiAllelicFrequency minimum frequency allowed for the first and second most common allele. Setting a Lower value for this parameter will minimise the identification of loci with three or more alleles in one sample. This is useful if sequencing errors are suspected to be common.
maxReads max number of reads of one list-element allowed
verbose logical indicating if process information should be displayed

Details

This function scans all reads stored in a GAlignmentsList for possible heterozygote positions. The user can balance the sensitivity of the search by modifying the minimumReadsAtPos, maximumMajorAlleleFrequency and minimumBiAllelicFrequency arguments.

Value

scanForHeterozygotes.old returns a GRanges object with the SNPs for the BamList object that was used as input.

Author(s)

Jesper R. Gadin, Lasse Folkersen

See Also

• The getAlleleCounts which is a function that count the number of reads overlapping a site.

Examples

data(reads)
s <- scanForHeterozygotes.old(reads, verbose=FALSE)
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