Package 'sveval'

March 5, 2019

Title SV evaluation

Version 1.2.0

Description Evaluate SV in a call set against a truth set using overlap-based approaches and sequence comparison for insertions.

Depends R (¿= 3.4.4)

License MIT + file LICENSE

Encoding UTF-8

LazyData true

RoxygenNote 6.1.1

Imports VariantAnnotation,
GenomicRanges,
IRanges,
magrittr,
dplyr,
rlang,
DelayedArray,
Biostrings,
parallel,
testthat,
ggplot2

R topics documented:

sveval-package *SV evaluation*

Description

Evaluate SV in a call set against a truth set using overlap-based approaches and sequence comparison for insertions.

Details

Package:	sveval
Type:	Package
Version:	1.2.0
Date:	2019-02-28
License:	MIT

Author(s)

Jean Monlong <jmonlong@ucsc.edu>

See Also

http://www.github.com/jmonlong/sveval

Examples

```
## Not run:
eval = svevalOl('calls.vcf', 'truth.vcf')
plot_prcurve(eval$curve)

# Comparing multiple methods
eval.1 = svevalOl('calls1.vcf', 'truth.vcf')
eval.2 = svevalOl('calls2.vcf', 'truth.vcf')
plot_prcurve(list(eval.1$curve, eval.2$curve), labels=c('method1', 'method2'))

## End(Not run)
```

filterSVs *Filter SVs for size and regions of interest*

Description

Filter SVs for size and regions of interest

Usage

```
filterSVs(sv.gr, regions.gr = NULL, ol.prop = 0.5, min.size = 0,
  max.size = Inf)
```

Arguments

sv.gr	the input SVs (e.g. read from readSVvcf)
regions.gr	the regions of interest. Ignored if NULL (default).
ol.prop	minimum proportion of sv.gr that must overlap regions.gr. Default is 0.5
min.size	the minimum SV size to be considered. Default 0.
max.size	the maximum SV size to be considered. Default is Inf.

Value

a subset of sv.gr that overlaps regions.gr or in the specified size range.

Author(s)

Jean Monlong

---

plot_prcurve *Create precision-recall graphs*

Description

Create a precision/recall curve using metrics computed by the svevalOl function. The svevalOl function returns a list containing a "curve" data.frame with the evaluation metrics for different quality thresholds.

Usage

```
plot_prcurve(eval, labels = NULL)
```

Arguments

eval	a data.frame, a list of data.frames, or a vector with one or several paths to files with "curve" information.
labels	the labels to use for each input (when multiple inputs are used). Ignored is NULL (default).

Details

If the input is a data.frame (or list of data.frames) it should be the "curve" element of the list returned by the svevalOl function. If the input is a character (or a vector of characters), they are considered to be file names and the data will be read from these files.

If multiple inputs are given, either using a list of data.frames or a vectors with several filenames, one curve per input will be created. This is to be used to quickly compare several methods. The "labels" parameters can be used to specify a label for each input to use for the graphs.

Value

list of ggplot graph objects

Author(s)

Jean Monlong

Examples

```
## Not run:
eval = svevalOl('calls.vcf', 'truth.vcf')
plot_prcurve(eval$curve)

# Comparing multiple methods
eval.1 = svevalOl('calls1.vcf', 'truth.vcf')
eval.2 = svevalOl('calls2.vcf', 'truth.vcf')
plot_prcurve(list(eval.1$curve, eval.2$curve), labels=c('method1', 'method2'))

# Or if the results were previously written in files
plot_prcurve(c('methods1-prcurve.tsv', 'methods2-prcurve.tsv'), labels=c('method1', 'method2'))

## End(Not run)
```

---

readSVvcf *Read SVs from a VCF file*

Description

Read a VCF file that contains SVs and create a GRanges with relevant information, e.g. SV size or genotype quality.

Usage

```
readSVvcf(vcf.file, keep.ins.seq = FALSE, sample.name = NULL,
  qual.field = c("QUAL", "GQ"), check.inv = FALSE)
```

Arguments

vcf.file	the path to the VCF file
keep.ins.seq	should it keep the inserted sequence? Default is FALSE.
sample.name	the name of the sample to use. If NULL (default), use first sample.
qual.field	fields to use as quality. Will be tried in order.
check.inv	should the sequence of MNV be compared to identify inversions.

Details

By default, the quality information is taken from the QUAL field. If all values are NA or 0, the function will try other fields as speficied in the "qual.field" vector. Fields can be from the INFO or FORMAT fields.

Value

a GRanges object with relevant information.

Author(s)

Jean Monlong

Examples

```
## Not run:
calls.gr = readSVvcf('calls.vcf')

## End(Not run)
```

`svevalOl` *SV evaluation based on overlap and variant size*

Description

SV evaluation based on overlap and variant size

Usage

```
svevalOl(calls.gr, truth.gr, max.ins.dist = 20, min.cov = 0.5,
  min.del.rol = 0.1, ins.seq.comp = FALSE, nb.cores = 1,
  min.size = 50, max.size = Inf, bed.regions = NULL,
  bed.regions.ol = 0.5, qual.field = c("QUAL", "GQ"),
  sample.name = NULL, outfile = NULL, out.bed.prefix = NULL,
  qual.quantiles = seq(0, 1, 0.1), check.inv = FALSE,
  geno.eval = FALSE, stitch.hets = FALSE, stitch.dist = 20,
  merge.hets = FALSE, merge.rol = 0.8)
```

Arguments

`calls.gr`	call set. A GRanges or the path to a VCF file.
`truth.gr`	truth set. A GRanges or the path to a VCF file.
`max.ins.dist`	maximum distance for insertions to be clustered. Default is 20.
`min.cov`	the minimum coverage to be considered a match. Default is 0.5
`min.del.rol`	minimum reciprocal overlap for deletions. Default is 0.1
`ins.seq.comp`	compare sequence instead of insertion sizes. Default is FALSE.
`nb.cores`	number of processors to use. Default is 1.
`min.size`	the minimum SV size to be considered. Default 50.
`max.size`	the maximum SV size to be considered. Default is Inf.
`bed.regions`	If non-NULL, a GRanges object or path to a BED file (no headers) with regions of interest.

bed.regions.ol	minimum proportion of sv.gr that must overlap regions.gr. Default is 0.5
qual.field	fields to use as quality. Will be tried in order.
sample.name	the name of the sample to use if VCF files given as input. If NULL (default), use first sample.
outfile	the TSV file to output the results. If NULL (default), returns a data.frame.
out.bed.prefix	prefix for the output BED files. If NULL (default), no BED output.
qual.quantiles	the QUAL quantiles for the PR curve. Default is (0, .1, ..., .9, 1).
check.inv	should the sequence of MNV be compared to identify inversions.
geno.eval	should het/hom be evaluated separately (genotype evaluation). Default FALSE.
stitch.hets	should clustered hets be stitched together before genotype evatuation. Default is FALSE.
stitch.dist	the maximum distance to stitch hets during genotype evaluation.
merge.hets	should similar hets be merged into homs before genotype evaluation. Default is FALSE.
merge.rol	the minimum reciprocal overlap to merge hets before genotype evaluation.

Value

a list with

eval	a data.frame with TP, FP and FN for each SV type when including all variants
curve	a data.frame with TP, FP and FN for each SV type when using different quality thesholds

Author(s)

Jean Monlong

Examples

```
## Not run:
## From VCF files
eval = svevalOl('calls.vcf', 'truth.vcf')

## From GRanges
calls.gr = readSVvcf('calls.vcf')
truth.gr = readSVvcf('truth.vcf')
eval = svevalOl(calls.gr, truth.gr)

## Genotype evaluation
eval = svevalOl(calls.gr, truth.gr, geno.eval=TRUE, merge.hets=TRUE, stitch.hets=TRUE)

## End(Not run)
```

Index