

Package ‘sveval’

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Title SV evaluation

Version 1.2.1

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)

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parallel,
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ggplot2

R topics documented:

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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: 1.2.1
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License: MIT

Author(s)

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See Also

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

Examples

```
## Not run:  
eval = sveval01('calls.vcf', 'truth.vcf')  
plot_prcurve(eval$curve)  
  
# Comparing multiple methods  
eval.1 = sveval01('calls1.vcf', 'truth.vcf')  
eval.2 = sveval01('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	<i>Create precision-recall graphs</i>
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Description

Create a precision/recall curve using metrics computed by the sveval01 function. The sveval01 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 sveval01 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 = sveval01('calls.vcf', 'truth.vcf')
plot_prcurve(eval$curve)

# Comparing multiple methods
eval.1 = sveval01('calls1.vcf', 'truth.vcf')
eval.2 = sveval01('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("GQ", "QUAL"), 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 specified 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)
```

sveval0l

SV evaluation based on overlap and variant size

Description

SV evaluation based on overlap and variant size

Usage

```
sveval0l(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.ths = c(0, 2, 3, 4, 5, 7, 10, 12, 14, 21, 27, 35, 45, 50, 60, 75,
  90, 99, 110, 133, 167, 180, 250, 350, 450, 550, 650),
  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

<code>calls.gr</code>	call set. A GRanges or the path to a VCF file.
<code>truth.gr</code>	truth set. A GRanges or the path to a VCF file.
<code>max.ins.dist</code>	maximum distance for insertions to be clustered. Default is 20.
<code>min.cov</code>	the minimum coverage to be considered a match. Default is 0.5
<code>min.del.rol</code>	minimum reciprocal overlap for deletions. Default is 0.1
<code>ins.seq.comp</code>	compare sequence instead of insertion sizes. Default is FALSE.
<code>nb.cores</code>	number of processors to use. Default is 1.
<code>min.size</code>	the minimum SV size to be considered. Default 50.
<code>max.size</code>	the maximum SV size to be considered. Default is Inf.

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

Value

a list with	
<code>eval</code>	a data.frame with TP, FP and FN for each SV type when including all variants
<code>curve</code>	a data.frame with TP, FP and FN for each SV type when using different quality thresholds
<code>svs</code>	a list of GRanges object with FP, TP and FN for each SV type (when using $QUAL_i=0$ threshold).

Author(s)

Jean Monlong

Examples

```
## Not run:
## From VCF files
eval = sveval01('calls.vcf', 'truth.vcf')

## From GRanges
calls.gr = readSVvcf('calls.vcf')
truth.gr = readSVvcf('truth.vcf')
```

```
eval = sveval01(calls.gr, truth.gr)

## Genotype evaluation
eval = sveval01(calls.gr, truth.gr, geno.eval=TRUE, merge.hets=TRUE, stitch.hets=TRUE)

## End(Not run)
```

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