Package 'sveval'

February 28, 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 ($i = 3.4.4$)		
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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: 1.2.0
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

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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

S	Create precision-recall g	plot_prcurve
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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.

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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

 ${\tt vcf.file} \qquad \qquad {\tt the~path~to~the~VCF~file}$

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.

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Value

a GRanges object with relevant information.

Author(s)

Jean Monlong

Examples

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

sveval01

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 = 0, max.size = Inf, bed.regions = NULL,
bed.regions.ol = 0.5, sample.name = NULL, outfile = NULL,
out.bed.prefix = NULL, qual.quantiles = seq(0, 1, 0.1),
check.inv = FALSE)
```

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.
                 maximum distance for insertions to be clustered. Default is 20.
max.ins.dist
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.
                 the minimum SV size to be considered. Default 0.
min.size
                 the maximum SV size to be considered. Default is Inf.
max.size
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
```

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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.

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 the sholds

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)
## End(Not run)
```

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