Package 'vcfppR'

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Title Rapid Manipulation of the Variant Call Format (VCF)

Version 0.8.0

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Description The 'vcfpp.h' (https://github.com/Zilong-Li/vcfpp) provides an easy-to-use 'C++' 'API' of 'htslib', offering full functionality for manipulating Variant Call Format (VCF) files. The 'vcfppR' package serves as the R bindings of the 'vcfpp.h' library, enabling rapid processing of both compressed and uncompressed VCF files. Explore a range of powerful features for efficient VCF data manipulation.

Encoding UTF-8

Depends R (>= 3.6.0)

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Suggests knitr, codetools, rmarkdown, testthat (>= 3.0.0)

Config/testthat/edition 3

 ${\bf System Requirements} \ \ libcurl: libcurl-devel\ (rpm)\ or$

libcurl4-openssl-dev (deb), GNU make.

Imports Rcpp, methods, stats, utils

LinkingTo Rcpp

URL https://github.com/Zilong-Li/vcfppR

BugReports https://github.com/Zilong-Li/vcfppR/issues

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VignetteBuilder knitr NeedsCompilation yes

Author Zilong Li [aut, cre] (ORCID: https://orcid.org/0000-0001-5859-2078),

Bonfield, James K and Marshall, John and Danecek, Petr and Li, Heng and Ohan, Valeriu and Whitwham, Andrew and Keane, Thomas and Davies,

Robert M [cph] (Authors of included htslib library)

Maintainer Zilong Li <zilong.dk@gmail.com>

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Description

The 'vcfpp.h' (https://github.com/Zilong-Li/vcfpp) provides an easy-to-use 'C++' 'API' of 'htslib', offering full functionality for manipulating Variant Call Format (VCF) files. The 'vcfppR' package serves as the R bindings of the 'vcfpp.h' library, enabling rapid processing of both compressed and uncompressed VCF files. Explore a range of powerful features for efficient VCF data manipulation.

Author(s)

Maintainer: Zilong Li <zilong.dk@gmail.com> (ORCID)

Other contributors:

• Bonfield, James K and Marshall, John and Danecek, Petr and Li, Heng and Ohan, Valeriu and Whitwham, Andrew and Keane, Thomas and Davies, Robert M (Authors of included htslib library) [copyright holder]

See Also

Useful links:

- https://github.com/Zilong-Li/vcfppR
- Report bugs at https://github.com/Zilong-Li/vcfppR/issues

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vcfcomp

Compare two VCF/BCF files reporting various statistics

Description

Compare two VCF/BCF files reporting various statistics

Usage

```
vcfcomp(
  test,
  truth,
  formats = c("DS", "GT"),
  stats = "r2",
  by.sample = FALSE,
  by.variant = FALSE,
  flip = FALSE,
  names = NULL,
  bins = NULL,
  out = NULL,
  choose_random_start = FALSE,
  return_pse_sites = FALSE,
  ...
)
```

Arguments

test

| | or saved RDS file. |
|------------|---|
| truth | path to the baseline file (truth), which can be a VCF/BCF file, vcftable object or saved RDS file. |
| formats | character vector. the FORMAT tags to extract for the test and truth respectively. default $c("DS", "GT")$ extracts 'DS' of the target and 'GT' of the truth. |
| stats | the statistics to be calculated. supports the following. "r2": the Pearson correlation coefficient square. "f1": the F1-score, good balance between sensitivity and precision. "nrc": the Non-Reference Concordance rate "pse": the Phasing Switch Error rate |
| by.sample | logical. calculate sample-wise concordance, which can be stratified by MAF bin. |
| by.variant | logical. calculate variant-wise concordance, which can be stratified by MAF bin. If both by.sample and by.variant are FALSE, then do calculations for all |

path to the comparision file (test), which can be a VCF/BCF file, vcftable object

flip logical. flip the ref and alt variants

names character vector. reset samples' names in the test VCF.

samples and variants together in a bin.

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bins numeric vector. break statistics into allele frequency bins.

af file path with allele frequency or a RDS file with a saved object for af. Format

of the text file: a space-separated text file with five columns and a header named

'chr' 'pos' 'ref' 'alt' 'af'

out output prefix for saving objects into RDS file

choose_random_start

choose random start for stats="pse"

return_pse_sites

boolean. return phasing switch error sites

... options passed to vcftable

Details

vcfcomp implements various statistics to compare two VCF/BCF files, e.g. report genotype concordance, correlation stratified by allele frequency.

Value

a list of various statistics

Author(s)

```
Zilong Li <zilong.dk@gmail.com>
```

Examples

```
library('vcfppR')
test <- system.file("extdata", "imputed.gt.vcf.gz", package="vcfppR")
truth <- system.file("extdata", "imputed.gt.vcf.gz", package="vcfppR")
samples <- "HG00133,HG00143,HG00262"
res <- vcfcomp(test, truth, stats="f1", samples=samples, setid=TRUE)
str(res)</pre>
```

vcfinfo

read a INFO tag in the VCF/BCF into R data structure

Description

read a INFO tag in the VCF/BCF into R data structure

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Usage

```
vcfinfo(
  vcffile,
  tag,
  region = "",
  vartype = "all",
  ids = NULL,
  qual = 0,
  pass = FALSE,
  setid = FALSE
)
```

Arguments

| vcffile | path to the VCF/BCF file |
|---------|--|
| tag | the INFO tag to extract. |
| region | region to subset in bcftools-like style: "chr1", "chr1:1-10000000" |
| vartype | $restrict \ to \ specific \ type \ of \ variants. \ supports \ "snps", "indels", "sv", "multisnps", "multiallelics"$ |
| ids | character vector. restrict to sites with ID in the given vector. default NULL won't filter any sites. |
| qual | numeric. restrict to variants with QUAL > qual. |
| pass | logical. restrict to variants with FILTER = "PASS". |
| setid | logical. reset ID column as CHR_POS_REF_ALT. |
| | |

Details

vcfinfo uses the C++ API of vcfpp, which is a wrapper of htslib, to read VCF/BCF files. Thus, it has the full functionalities of htslib, such as restrict to specific variant types, samples and regions. For the memory efficiency reason, the vcfinfo is designed to parse only one tag at a time in the INFO column of the VCF. Currently it does not support parsing a vector of values for a given INFO tag.

Value

Return a list containing the following components:

chr : character vector;

the CHR column in the VCF file

pos : character vector;

the POS column in the VCF file

id : character vector;

the ID column in the VCF file

ref: character vector;

the REF column in the VCF file

alt : character vector;

the ALT column in the VCF file

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```
qual: character vector;
```

the QUAL column in the VCF file

filter: character vector;

the FILTER column in the VCF file

tag: vector of either integer, numberic or character values depending on the tag to extract;

a specifiy tag in the INFO column to be extracted

Author(s)

```
Zilong Li <zilong.dk@gmail.com>
```

Examples

```
library('vcfppR')
vcffile <- system.file("extdata", "raw.gt.vcf.gz", package="vcfppR")
res <- vcfinfo(vcffile, "AF", region = "chr21:1-5050000", vartype = "snps", pass = TRUE)
str(res)</pre>
```

vcfplot

Make sensible and beautiful plots based on various objects in vcfppR

Description

Make sensible and beautiful plots based on various objects in vcfppR

Usage

```
vcfplot(obj, which.sample = NULL, variant = c("SNP", "INDEL"), pop = NULL, ...)
```

Arguments

obj object returned by vcftable, vcfcomp, vcfsummary

which sample which sample to be plotted. NULL will aggregate all samples.

variant which types of variant are desired pop file contains population information

... parameters passed to graphics

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vcfpopgen

count the heterozygous sites per sample in the VCF/BCF

Description

count the heterozygous sites per sample in the VCF/BCF

Usage

```
vcfpopgen(
  vcffile,
  region = "",
  samples = "-",
  pass = FALSE,
  qual = 0,
  fun = "heterozygosity"
)
```

Arguments

```
vcffile path to the VCF/BCF file
region region to subset like bcftools
samples samples to subset like bcftools
pass restrict to variants with FILTER==PASS
qual restrict to variants with QUAL > qual.
```

which popgen function to run. available functions are "heterozygosity".

Value

fun

vcfpopgen a list containing the following components:

samples: character vector;

the samples ids in the VCF file after subsetting

hets: integer vector;

the counts of heterozygous sites of each sample in the same order as samples

Author(s)

```
Zilong Li <zilong.dk@gmail.com>
```

Examples

```
library('vcfppR')
vcffile <- system.file("extdata", "raw.gt.vcf.gz", package="vcfppR")
res <- vcfpopgen(vcffile)
str(res)</pre>
```

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vcfreader

API for manipulating the VCF/BCF.

Description

Type the name of the class to see the details and methods

Value

A C++ class with the following fields/methods for manipulating the VCF/BCF

Fields

new Constructor given a vcf file

• Parameter: vcffile - The path of a vcf file

new Constructor given a vcf file and the region

- Parameter: vcffile The path of a vcf file
- Parameter: region The region to be constrained

new Constructor given a vcf file, the region and the samples

- Parameter: vcffile The path of a vcf file
- Parameter: region The region to be constrained
- Parameter: samples The samples to be constrained. Comma separated list of samples to include (or exclude with "^" prefix).

setRegion try to set specific region to work with. will throw errors if no index or region found. Use getStatus to check if the region is valid or empty!

getStatus return 1: region is valid and not empty. 0: region is valid but empty. -1: no index file. -2: region not found or invalid region form

variant Try to get next variant record. return FALSE if there are no more variants or hit the end of file, otherwise TRUE.

chr Return the CHROM field of current variant

pos Return the POS field of current variant

id Return the CHROM field of current variant

ref Return the REF field of current variant

alt Return the ALT field of current variant

qual Return the QUAL field of current variant

filter Return the FILTER field of current variant

info Return the INFO field of current variant

infoInt Return the tag value of integer type in INFO field of current variant

• Parameter: tag - The tag name to retrieve in INFO

infoFloat Return the tag value of float type in INFO field of current variant

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• Parameter: tag - The tag name to retrieve in INFO

infoStr Return the tag value of string type in INFO field of current variant

• Parameter: tag - The tag name to retrieve in INFO

infoIntVec Return the tag value in a vector of integer type in INFO field of current variant

• Parameter: tag - The tag name to retrieve in INFO

infoFloatVec Return the tag value in a vector of float type in INFO field of current variant

• Parameter: tag - The tag name to retrieve in INFO

genotypes Return the genotype values in a vector of integers

• Parameter: collapse - Boolean value indicates wheather to collapse the size of genotypes, eg, return diploid genotypes.

formatInt Return the tag value of integer type for each sample in FORAMT field of current variant

• Parameter: tag - The tag name to retrieve in FORAMT

formatFloat Return the tag value of float type for each sample in FORAMT field of current variant

• Parameter: tag - The tag name to retrieve in FORAMT

formatStr Return the tag value of string type for each sample in FORAMT field of current variant

• Parameter: tag - The tag name to retrieve in FORAMT

isSNP Test if current variant is exculsively a SNP or not

isIndel Test if current variant is exculsively a INDEL or not

isSV Test if current variant is exculsively a SV or not

isMultiAllelics Test if current variant is exculsively a Multi Allelics or not

isMultiAllelicSNP Test if current variant is exculsively a Multi Biallelics (SNPs) or not

has SNP Test if current variant has a SNP or not

has INDEL Test if current variant has a INDEL or not

has INS Test if current variant has a INS or not

hasDEL Test if current variant has a DEL or not

hasMNP Test if current variant has a MNP or not

hasBND Test if current variant has a BND or not

hasOTHER Test if current variant has a OTHER or not

hasOVERLAP Test if current variant has a OVERLAP or not

nsamples Return the number of samples

samples Return a vector of samples id

header Return the raw string of the vcf header

string Return the raw string of current variant including newline

line Return the raw string of current variant without newline

output Init an output object for streaming out the variants to another vcf

updateSamples update samples name in the output VCF

• Parameter: s - A comma-seperated string for new samples names

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write Streaming out current variant the output vcf

close Close the connection to the output vcf

setCHR Modify the CHR of current variant

• Parameter: s - A string for CHR

setID Modify the ID of current variant

• Parameter: s - A string for ID

setPOS Modify the POS of current variant

• Parameter: pos - An integer for POS

setRefAlt Modify the REF and ALT of current variant

• Parameter: s - A string reperated by comma

setInfoInt Modify the given tag of INT type in the INFO of current variant

- Parameter: tag A string for the tag name
- Parameter: v An integer for the tag value

setInfoFloat Modify the given tag of FLOAT type in the INFO of current variant

- Parameter: tag A string for the tag name
- Parameter: v A double for the tag value

setInfoStr Modify the given tag of STRING type in the INFO of current variant

- Parameter: tag A string for the tag name
- Parameter: s A string for the tag value

setPhasing Modify the phasing status of each sample

• Parameter: v - An integer vector with size of the number of samples. only 1s and 0s are valid.

setGenotypes Modify the genotypes of current variant

• Parameter: v - An integer vector for genotypes. Use NA or -9 for missing value.

setFormatInt Modify the given tag of INT type in the FORMAT of current variant

- Parameter: tag A string for the tag name
- Parameter: v An integer for the tag value

setFormatFloat Modify the given tag of FLOAT type in the FORMAT of current variant

- Parameter: tag A string for the tag name
- Parameter: v A double for the tag value

setFormatStr Modify the given tag of STRING type in the FORMAT of current variant

- Parameter: tag A string for the tag name
- Parameter: s A string for the tag value

rmInfoTag Remove the given tag from the INFO of current variant

• Parameter: s - A string for the tag name

rmFormatTag Remove the given tag from the FORMAT of current variant

• Parameter: s - A string for the tag name

setVariant Modify current variant by adding a vcf line

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• Parameter: s - A string for one line in the VCF

addINFO Add a INFO in the header of the vcf

- Parameter: id A string for the tag name
- Parameter: number A string for the number
- Parameter: type A string for the type
- Parameter: desc A string for description of what it means

addFORMAT Add a FORMAT in the header of the vcf

- Parameter: id A string for the tag name
- Parameter: number A string for the number
- Parameter: type A string for the type
- Parameter: desc A string for description of what it means

Examples

```
vcffile <- system.file("extdata", "raw.gt.vcf.gz", package="vcfppR")
br <- vcfreader$new(vcffile)
res <- rep(0L, br$nsamples())
while(br$variant()) {
  if(br$isSNP()) {
   gt <- br$genotypes(TRUE) == 1
   gt[is.na(gt)] <- FALSE
   res <- res + gt
  }
}</pre>
```

vcfsummary

summarize the various variant types at both variant level and sample level.

Description

summarize the various variant types at both variant level and sample level.

Usage

```
vcfsummary(
  vcffile,
  region = "",
  samples = "-",
  pass = FALSE,
  qual = 0,
  svtype = FALSE
)
```

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Arguments

vcffile path to the VCF/BCF file
region region to subset like bcftools
samples samples to subset like bcftools
pass restrict to variants with FILTER==PASS
qual restrict to variants with QUAL > qual.
svtype summarize the variants with SVTYPE

Details

```
bcftools view -s "id01,id02" input.bcf.gz chr1:100000-20000
```

Value

vcfsummary a list containing the following components:

summary : named integer vector;

summarize the counts of each variant type

samples: character vector;

the samples ids in the VCF file after subsetting

vartype: integer vector;

the counts of the variant type at sample level in the same order as samples

Author(s)

```
Zilong Li <zilong.dk@gmail.com>
```

Examples

```
library('vcfppR')
svfile <- system.file("extdata", "sv.vcf.gz", package="vcfppR")
res <- vcfsummary(svfile, region = "chr21:1-10000000", svtype = TRUE)
str(res)</pre>
```

vcftable

read VCF/BCF contents into R data structure

Description

The swiss army knife for reading VCF/BCF into R data types rapidly and easily.

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Usage

```
vcftable(
  vcffile,
  region = "",
  samples = "-",
  vartype = "all",
  format = "GT",
  ids = NULL,
  qual = 0,
  pass = FALSE,
  info = TRUE,
  collapse = TRUE,
  setid = FALSE,
  mac = 0,
  rmdup = FALSE
)
```

Arguments

vcffile path to the VCF/BCF file

region region to subset in bcftools-like style: "chr1", "chr1:1-10000000"

samples samples to subset in beftools-like style. comma separated list of samples to

include (or exclude with "^" prefix). e.g. "id01,id02", "^id01,id02".

vartype restrict to specific type of variants. supports "snps", "indels", "sv", "multisnps", "multiallelics"

format the FORMAT tag to extract. default "GT" is extracted.

ids character vector. restrict to sites with ID in the given vector. default NULL

won't filter any sites.

qual numeric. restrict to variants with QUAL > qual.

pass logical. restrict to variants with FILTER = "PASS".

info logical. drop INFO column in the returned list.

collapse logical. It acts on the FORMAT. If the FORMAT to extract is "GT", the dim of

raw genotypes matrix of diploid is (M, 2 * N), where M is #markers and N is #samples. default TRUE will collapse the genotypes for each sample such that the matrix is (M, N). Set this to FALSE if one wants to maintain the phasing order, e.g. "110" is parsed as c(1, 0) with collapse=FALSE. If the FORMAT to extract is not "GT", then with collapse=TRUE it will try to turn a list of the extracted vector into a matrix. However, this raises issues when one variant is

mutliallelic resulting in more vaules than others.

setid logical. reset ID column as CHR_POS_REF_ALT.

mac integer. restrict to variants with minor allele count higher than the value.

rmdup logical. remove duplicated sites by keeping the first occurrence of POS. (default:

FALSE)

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Details

vcftable uses the C++ API of vcfpp, which is a wrapper of htslib, to read VCF/BCF files. Thus, it has the full functionalities of htslib, such as restrict to specific variant types, samples and regions. For the memory efficiency reason, the vcftable is designed to parse only one tag at a time in the FORMAT column of the VCF. In default, only the matrix of genotypes, i.e. "GT" tag, are returned by vcftable, but there are many other tags supported by the format option.

Value

Return a list containing the following components:

samples : character vector;

the samples ids in the VCF file after subsetting

chr: character vector;

the CHR column in the VCF file

pos: character vector;

the POS column in the VCF file

id: character vector;

the ID column in the VCF file

ref: character vector;

the REF column in the VCF file

alt : character vector;

the ALT column in the VCF file

qual: character vector;

the QUAL column in the VCF file

filter: character vector;

the FILTER column in the VCF file

info : character vector;

the INFO column in the VCF file

format: matrix of either integer or numberic values depending on the tag to extract; a specify tag in the FORMAT column to be extracted

Author(s)

```
Zilong Li <zilong.dk@gmail.com>
```

Examples

```
library('vcfppR')
vcffile <- system.file("extdata", "raw.gt.vcf.gz", package="vcfppR")
res <- vcftable(vcffile, "chr21:1-5050000", vartype = "snps")
str(res)</pre>
```

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vcfwriter

API for writing the VCF/BCF.

Description

Type the name of the class to see the details and methods

Value

A C++ class with the following fields/methods for writing the VCF/BCF

Fields

new Constructor given a vcf file

- Parameter: vcffile The path of a vcf file. don't start with "~"
- Parameter: version The version of VCF specification

addContig Add a Contig in the header of the vcf

• Parameter: str - A string for the CONTIG name

addFILTER Add a FILTER in the header of the vcf

- Parameter: id A string for the FILTER name
- Parameter: desc A string for description of what it means

addINFO Add a INFO in the header of the vcf

- Parameter: id A string for the tag name
- Parameter: number A string for the number
- Parameter: type A string for the type
- Parameter: desc A string for description of what it means

addFORMAT Add a FORMAT in the header of the vcf

- Parameter: id A string for the tag name
- Parameter: number A string for the number
- Parameter: type A string for the type
- Parameter: desc A string for description of what it means

addSample Add a SAMPLE in the header of the vcf

• Parameter: str - A string for a SAMPLE name

addLine Add a line in the header of the vcf

• Parameter: str - A string for a line in the header of VCF

writeline Write a variant record given a line

• Parameter: line - A string for a line in the variant of VCF. Not ended with "newline"

close Close and save the vcf file

vefwriter vefwriter

Examples

```
outvcf <- file.path(paste0(tempfile(), ".vcf.gz"))
bw <- vcfwriter$new(outvcf, "VCF4.1")
bw$addContig("chr20")
bw$addFORMAT("GT", "1", "String", "Genotype");
bw$addSample("NA12878")
s1 <- "chr20\t20060600\t.\tG\tC\t100\tPASS\t.\tGT\t1|0"
bw$writeline(s1)
bw$close()</pre>
```

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