RBcf: An VCF API for R.

Pierre Lindenbaum / yokofakun/ Institut du Thorax . Nantes. ${\it April~23,~2020}$

1 Abstract

RBcf uses the Htslib C API for parsing VCF and BCF files. This API was written by a regular user of the htsjdk library who doesn't like R.

2 Examples

2.1 Open and close a VCF file

Code:

```
# load rbcf
library(rbcf)
# we don't need the index for this file
fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)
# dispose the vcf reader
bcf.close(fp)

Output:
> # load rbcf
> library(rbcf)
> # we don't need the index for this file
> fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
```

2.2Print the INFOs in the VCF header

```
Code:
```

```
# load rbcf
library(rbcf)
# we don't need the index for this file
fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
info <- bcf.infos(fp)</pre>
# dispose the vcf reader
bcf.close(fp)
# print the table
info
Output:
> # load rbcf
> library(rbcf)
> # we don't need the index for this file
> fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
> info <- bcf.infos(fp)</pre>
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
> # print the table
> info
         ID Number
                        Type
INDEL INDEL
                  0
                       Flag
IDV
                  1 Integer
        IDV
IMF
        IMF
                  1
                      Float
DP
         DP
                  1 Integer
        VDB
                      Float
VDB
                  1
RPB
        RPB
                  1
                      Float
```

```
MQB
        MQB
                  1
                      Float
                  1
BQB
        BQB
                      Float
       MQSB
                      Float
MQSB
                  1
SGB
        SGB
                  1
                      Float
MQOF
       MQOF
                      Float
                  1
ICB
                      Float
        ICB
                  1
HOB
        HOB
                       Float
                  1
AC
          AC
                  A Integer
AN
          AN
                  1 Integer
```

```
DP4
        DP4
                 4 Integer
MQ
         MQ
                 1 Integer
INDEL
                                                             "Indicates that the v
TDV
                                                         "Maximum number of reads
                                                       "Maximum fraction of reads
IMF
DΡ
      "Variant Distance Bias for filtering splice-site artefacts in RNA-seq data
VDB
RPB
                                       "Mann-Whitney U test of Read Position Bias
MQB
                                     "Mann-Whitney U test of Mapping Quality Bias
BQB
                                        "Mann-Whitney U test of Base Quality Bias
MQSB
                           "Mann-Whitney U test of Mapping Quality vs Strand Bias
SGB
                                                                           "Segreg
MQOF
                                                           "Fraction of MQO reads
                                            "Inbreeding Coefficient Binomial test
ICB
HOB
                                              "Bias in the number of HOMs number
                        "Allele count in genotypes for each ALT allele, in the s
AC
AN
                                                         "Total number of alleles
DP4
               "Number of high-quality ref-forward , ref-reverse, alt-forward an
MQ
                                                                             "Aver
>
```

2.3 Print the FORMATs in the VCF header

Code:

```
# load rbcf
library(rbcf)
# we don't need the index for this file
fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)
fmts <- bcf.formats(fp)
# dispose the vcf reader
bcf.close(fp)
# print the table
fmts
Output:
> # load rbcf
> library(rbcf)
> # we don't need the index for this file
> fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
```

```
> fmts <- bcf.formats(fp)</pre>
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
> # print the table
> fmts
   ID Number
                Туре
                                                        Description
PL PL
           G Integer "List of Phred-scaled genotype likelihoods"
GT GT
           1 String
                                                         "Genotype"
>
      Print the FILTERs in the VCF header
Code:
# load rbcf
library(rbcf)
# we don't need the index for this file
fp <- bcf.open("../tests/data/gnomad.exomes.r2.0.1.sites.vcf",FALSE)</pre>
flt <- bcf.filters(fp)</pre>
# dispose the vcf reader
bcf.close(fp)
# print the table
flt
Output:
> # load rbcf
> library(rbcf)
> # we don't need the index for this file
> fp <- bcf.open("../tests/data/gnomad.exomes.r2.0.1.sites.vcf",FALSE)</pre>
> flt <- bcf.filters(fp)</pre>
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
> # print the table
> flt
                              ID
PASS
                            PASS
AC0
                             AC0
InbreedingCoeff InbreedingCoeff
```

LCR

LCR

```
RF
                              RF
                          SEGDUP
SEGDUP
PASS
AC0
                "Allele Count is zero (i.e. no high-confidence genotype (GQ >= 2
InbreedingCoeff
LCR
RF
                                                      "Failed random forests filte
SEGDUP
2.5
      Print the Samples in the VCF header
Code:
# load rbcf
library(rbcf)
# we don't need the index for this file
fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
# print the number of samples
cat(paste("Num. Samples=",bcf.nsamples(fp),".\n"))
# get the name for the 1st sample
cat(paste("First sample is ",bcf.sample1(fp,1),".\n"))
# get the samples
samples <- bcf.samples(fp)</pre>
# dispose the vcf reader
bcf.close(fp)
# print the list
samples
Output:
> # load rbcf
> library(rbcf)
> # we don't need the index for this file
> fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
> # print the number of samples
> cat(paste("Num. Samples=",bcf.nsamples(fp),".\n"))
Num. Samples= 5 .
> # get the name for the 1st sample
> cat(paste("First sample is ",bcf.sample1(fp,1),".\n"))
```

```
First sample is S1.
> # get the samples
> samples <- bcf.samples(fp)
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
> # print the list
> samples
[1] "S1" "S2" "S3" "S4" "S5"
      Print the Dictionary in the VCF header
2.6
Code:
# load rbcf
library(rbcf)
# we don't need the index for this file
fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
dict <- bcf.dictionary(fp)</pre>
# dispose the vcf reader
bcf.close(fp)
# print the table
dict
Output:
> # load rbcf
> library(rbcf)
> # we don't need the index for this file
> fp <- bcf.open("../tests/data/rotavirus_rf.01.vcf",FALSE)</pre>
> dict <- bcf.dictionary(fp)</pre>
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
> # print the table
> dict
```

chrom size

RF01 RF01 3302 RF02 RF02 2687 RF03 RF03 2592

```
RF04 RF04 2362
RF05 RF05 1579
RF06 RF06 1356
RF07 RF07 1074
RF08 RF08 1059
RF09 RF09 1062
RF10 RF10 751
RF11 RF11 666
```

2.7 Print the Indexed Chromosomes

Code:

```
# load rbcf
library(rbcf)
# Open the indexed VCF
fp <- bcf.open("../tests/data/rotavirus_rf.02.vcf.gz")</pre>
# get the indexed contigs
contigs <- bcf.contigs(fp)</pre>
# dispose the vcf reader
bcf.close(fp)
# print the table
contigs
Output:
> # load rbcf
> library(rbcf)
> # Open the indexed VCF
> fp <- bcf.open("../tests/data/rotavirus_rf.02.vcf.gz")</pre>
> # get the indexed contigs
> contigs <- bcf.contigs(fp)</pre>
> # dispose the vcf reader
> bcf.close(fp)
[1] TRUE
> # print the table
> contigs
 [1] "RF01" "RF02" "RF03" "RF04" "RF05" "RF06" "RF07" "RF08" "RF09" "RF10"
[11] "RF11"
>
```