

An Introduction to *GenomeInfoDb*

Martin Morgan, Hervé Pagès, Marc Carlson, Sonali Arora

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1 Introduction

The *GenomeInfoDb* provides an interface to access seqlevelsStyles (such as UCSC, NCBI, Ensembl) and their supported mappings for organisms. For instance, for Homo sapiens, seqlevelsStyle "UCSC" maps to "chr1", "chr2", ..., "chrX", "chrY". The section below introduces these functions with examples.

2 Functionality for all existing organisms

2.1 genomeStyles

The genomeStyles lists out for each organism, the seqlevelsStyles and their mappings.

```
seqmap <- genomeStyles()
head(seqmap,n=2)

## $Arabidopsis_thaliana
##   circular auto sex NCBI TAIR9 Ensembl
## 1   FALSE TRUE FALSE 1 Chr1 1
## 2   FALSE TRUE FALSE 2 Chr2 2
## 3   FALSE TRUE FALSE 3 Chr3 3
## 4   FALSE TRUE FALSE 4 Chr4 4
## 5   FALSE TRUE FALSE 5 Chr5 5
## 6    TRUE FALSE FALSE MT ChrM Mt
## 7    TRUE FALSE TRUE Pltd ChrC Pt
##
## $Caenorhabditis_elegans
##   circular auto sex NCBI UCSC Ensembl
## 1   FALSE TRUE FALSE I chrI I
## 2   FALSE TRUE FALSE II chrII II
## 3   FALSE TRUE FALSE III chrIII III
## 4   FALSE TRUE FALSE IV chrIV IV
## 5   FALSE TRUE FALSE V chrV V
## 6   FALSE FALSE TRUE X chrX X
## 7    TRUE TRUE FALSE MT chrM MtDNA
```

Organism's supported by GenomeInfoDb can be found by :

```
names(genomeStyles())

## [1] "Arabidopsis_thaliana" "Caenorhabditis_elegans" "Canis_familiaris"
## [4] "Cyanidioschyzon_merolae" "Drosophila_melanogaster" "Homo_sapiens"
## [7] "Mus_musculus" "Oryza_sativa" "Populus_trichocarpa"
## [10] "Rattus_norvegicus" "Saccharomyces_cerevisiae" "Zea_mays"
## [13] "genomeMappingTbl.csv"
```

If one knows the organism one is interested in, then we can directly access the information for the given organism along. Each function accepts an argument called `species` which as "genus species", the default is "Homo sapiens". In the following example we list out only the first five entries returned by the code snippet.

```
head(genomeStyles("Homo_sapiens"),5)

##   circular auto sex NCBI UCSC dbSNP Ensembl
## 1   FALSE TRUE FALSE 1 chr1 ch1 1
## 2   FALSE TRUE FALSE 2 chr2 ch2 2
## 3   FALSE TRUE FALSE 3 chr3 ch3 3
## 4   FALSE TRUE FALSE 4 chr4 ch4 4
## 5   FALSE TRUE FALSE 5 chr5 ch5 5
```

We can also check if a given style is supported by GenomeInfoDb for a given species. For example, if we want to know if "UCSC" mapping is supported for "Homo sapiens" we can ask :

```
"UCSC" %in% names(genomeStyles("Homo_sapiens"))

## [1] TRUE
```

2.2 extractSeqlevels

We can also extract the desired `seqlevelsStyle` from a given organism using the `extractSeqlevels`

```
extractSeqlevels(species="Arabidopsis_thaliana", style="NCBI")
## [1] "1" "2" "3" "4" "5" "MT" "Pltd"
```

2.3 extractSeqlevelsByGroup

We can also extract the desired seqlevelsStyle from a given organism based on a group (Group - 'auto' denotes autosomes, 'circular' denotes circular chromosomes and 'sex' denotes sex chromosomes; the default is all chromosomes are returned).

```
extractSeqlevelsByGroup(species="Arabidopsis_thaliana", style="NCBI",
                        group="auto")
## [1] "1" "2" "3" "4" "5"
```

2.4 seqlevelsStyle

We can find the seqname Style for a given character vector by using the seqlevelsStyle

```
seqlevelsStyle(paste0("chr",c(1:30)))
## [1] "UCSC"
seqlevelsStyle(c("2L", "2R", "X", "Xhet"))
## [1] "NCBI"
```

2.5 seqlevelsInGroup

We can also subset a given character vector containing seqnames using the seqlevelsInGroup. We currently support 3 groups: 'auto' for autosomes, 'sex' for allosomes/sex chromosomes and circular for 'circular' chromosomes. The user can also provide the style and species they are working with. In the following examples, we extract the sex, auto and circular chromosomes for Homo sapiens :

```
newchr <- paste0("chr",c(1:22,"X","Y","M","1_g1000192_random","4_ctg9_hap1"))
seqlevelsInGroup(newchr, group="sex")
## [1] "chrX" "chrY"
seqlevelsInGroup(newchr, group="auto")
## [1] "chr1" "chr2" "chr3" "chr4" "chr5" "chr6" "chr7" "chr8" "chr9" "chr10"
## [11] "chr11" "chr12" "chr13" "chr14" "chr15" "chr16" "chr17" "chr18" "chr19" "chr20"
## [21] "chr21" "chr22"
seqlevelsInGroup(newchr, group="circular")
## [1] "chrM"
seqlevelsInGroup(newchr, group="sex", "Homo_sapiens", "UCSC")
## [1] "chrX" "chrY"
```

if we have a vector containing seqnames and we want to verify the species and style for them , we can use:

```
seqnames <- c("chr1", "chr9", "chr2", "chr3", "chr10")
all(seqnames %in% extractSeqlevels("Homo_sapiens", "UCSC"))
## [1] TRUE
```

2.6 orderSeqlevels

The `orderSeqlevels` can return the order of a given character vector which contains seqnames. In the following example, we show how you can find the order for a given seqnames character vector.

```
seqnames <- c("chr1", "chr9", "chr2", "chr3", "chr10")
orderSeqlevels(seqnames)

## [1] 1 3 4 2 5

seqnames[orderSeqlevels(seqnames)]

## [1] "chr1" "chr2" "chr3" "chr9" "chr10"
```

2.7 rankSeqlevels

The `rankSeqlevels` can return the rank of a given character vector which contains seqnames. In the following example, we show how you can find the rank for a given seqnames character vector.

```
seqnames <- c("chr1", "chr9", "chr2", "chr3", "chr10")
rankSeqlevels(seqnames)

## [1] 1 4 2 3 5
```

2.8 mapSeqlevels

Returns a matrix with 1 column per supplied sequence name and 1 row per sequence renaming map compatible with the specified style. If `best.only` is `TRUE` (the default), only the "best" renaming maps (i.e. the rows with less NAs) are returned.

```
mapSeqlevels(c("chrII", "chrIII", "chrM"), "NCBI")

## chrII chrIII chrM
## "II" "III" "MT"
```

We also have several seqlevel utility functions. Let us construct a basic `GRanges` and show how these functions can be used. .

```
gr <- GRanges(paste0("ch", 1:35), IRanges(1:35, width=5))
gr

## GRanges object with 35 ranges and 0 metadata columns:
##      seqnames      ranges strand
##      <Rle> <IRanges> <Rle>
##      [1]      ch1      [1, 5]      *
##      [2]      ch2      [2, 6]      *
##      [3]      ch3      [3, 7]      *
##      [4]      ch4      [4, 8]      *
##      [5]      ch5      [5, 9]      *
##      ...      ...      ...      ...
##      [31]     ch31     [31, 35]      *
##      [32]     ch32     [32, 36]      *
##      [33]     ch33     [33, 37]      *
##      [34]     ch34     [34, 38]      *
##      [35]     ch35     [35, 39]      *
##      -----
##      seqinfo: 35 sequences from an unspecified genome; no seqlengths
```

As you can see, we have "ch" instead of "chr" for chromosome names. We can use `renameSeqlevels` to change the "ch" to "chr"

2.9 renameSeqlevels

As the first argument - it takes the object whose seqlevels we need to change, and as the second argument it takes a named vector which has the changes.

```
newnames <- paste0("chr",1:35)
names(newnames) <- paste0("ch",1:35)
head(newnames)

##      ch1      ch2      ch3      ch4      ch5      ch6
## "chr1" "chr2" "chr3" "chr4" "chr5" "chr6"

gr <- renameSeqlevels(gr,newnames)
gr

## GRanges object with 35 ranges and 0 metadata columns:
##      seqnames      ranges strand
##      <Rle> <IRanges> <Rle>
##      [1]      chr1      [1, 5]      *
##      [2]      chr2      [2, 6]      *
##      [3]      chr3      [3, 7]      *
##      [4]      chr4      [4, 8]      *
##      [5]      chr5      [5, 9]      *
##      ...      ...      ...      ...
##      [31]     chr31     [31, 35]     *
##      [32]     chr32     [32, 36]     *
##      [33]     chr33     [33, 37]     *
##      [34]     chr34     [34, 38]     *
##      [35]     chr35     [35, 39]     *
##      -----
##      seqinfo: 35 sequences from an unspecified genome; no seqlengths
```

Humans have just 22 primary chromosomes - but here we have some extra seqlevels which we want to remove - there are several ways we can achieve this:

2.10 dropSeqlevels

Here the second argument is the seqlevels that you want to drop.

```
dropSeqlevels(gr,paste0("chr",23:35))

## GRanges object with 22 ranges and 0 metadata columns:
##      seqnames      ranges strand
##      <Rle> <IRanges> <Rle>
##      [1]      chr1      [1, 5]      *
##      [2]      chr2      [2, 6]      *
##      [3]      chr3      [3, 7]      *
##      [4]      chr4      [4, 8]      *
##      [5]      chr5      [5, 9]      *
##      ...      ...      ...      ...
##      [18]     chr18     [18, 22]     *
##      [19]     chr19     [19, 23]     *
```

```
## [20] chr20 [20, 24] *
## [21] chr21 [21, 25] *
## [22] chr22 [22, 26] *
## -----
## seqinfo: 22 sequences from an unspecified genome; no seqlengths
```

2.11 keepSeqlevels

Here the second argument is the seqlevels that you want to keep.

```
keepSeqlevels(gr, paste0("chr",1:22))

## GRanges object with 22 ranges and 0 metadata columns:
##      seqnames      ranges strand
##      <Rle> <IRanges> <Rle>
## [1] chr1      [1, 5]      *
## [2] chr2      [2, 6]      *
## [3] chr3      [3, 7]      *
## [4] chr4      [4, 8]      *
## [5] chr5      [5, 9]      *
## ...      ...      ...      ...
## [18] chr18     [18, 22]     *
## [19] chr19     [19, 23]     *
## [20] chr20     [20, 24]     *
## [21] chr21     [21, 25]     *
## [22] chr22     [22, 26]     *
## -----
## seqinfo: 22 sequences from an unspecified genome; no seqlengths
```

2.12 keepStandardChromosomes

This function internally uses the pre-defined tables inside *GenomeInfoDb* to find the correct seqlevels according to the style of the object.

```
keepStandardChromosomes(gr)

## GRanges object with 35 ranges and 0 metadata columns:
##      seqnames      ranges strand
##      <Rle> <IRanges> <Rle>
## [1] chr1      [1, 5]      *
## [2] chr2      [2, 6]      *
## [3] chr3      [3, 7]      *
## [4] chr4      [4, 8]      *
## [5] chr5      [5, 9]      *
## ...      ...      ...      ...
## [31] chr31     [31, 35]     *
## [32] chr32     [32, 36]     *
## [33] chr33     [33, 37]     *
## [34] chr34     [34, 38]     *
## [35] chr35     [35, 39]     *
## -----
## seqinfo: 35 sequences from an unspecified genome; no seqlengths
```

One can also specify the optional species argument to be more precise.

```

plantgr <- GRanges(c(1:5,"MT","Pltd"), IRanges(1:7,width=5))
keepStandardChromosomes(plantgr, species="Arabidopsis thaliana")

## GRanges object with 7 ranges and 0 metadata columns:
##      seqnames      ranges strand
##      <Rle> <IRanges>  <Rle>
## [1]      1      [1,  5]      *
## [2]      2      [2,  6]      *
## [3]      3      [3,  7]      *
## [4]      4      [4,  8]      *
## [5]      5      [5,  9]      *
## [6]      MT      [6, 10]      *
## [7]     Pltd      [7, 11]      *
## -----
## seqinfo: 7 sequences from an unspecified genome; no seqlengths

```

3 Classes inside GenomeInfoDb package

3.1 Genome-Description class

We also provide a Genome Description class which can be used in the following way:

```

library(BSgenome.Celegans.UCSC.ce2)
class(Celegans)

## [1] "BSgenome"
## attr("package")
## [1] "BSgenome"

is(Celegans, "GenomeDescription")

## [1] TRUE

provider(Celegans)

## [1] "UCSC"

seqinfo(Celegans)

## Seqinfo object with 7 sequences (1 circular) from ce2 genome:
##   seqnames seqlengths isCircular genome
##   chrI      15080483      FALSE    ce2
##   chrII      15279308      FALSE    ce2
##   chrIII     13783313      FALSE    ce2
##   chrIV      17493791      FALSE    ce2
##   chrV       20922231      FALSE    ce2
##   chrX       17718849      FALSE    ce2
##   chrM        13794         TRUE     ce2

gendesc <- as(Celegans, "GenomeDescription")
class(gendesc)

## [1] "GenomeDescription"
## attr("package")
## [1] "GenomeInfoDb"

gendesc

```

```
## | organism: Caenorhabditis elegans (Worm)
## | provider: UCSC
## | provider version: ce2
## | release date: Mar. 2004
## | release name: WormBase v. WS120
## | ---
## | seqlengths:
## |      chrI      chrII      chrIII      chrIV      chrV      chrX      chrM
## | 15080483 15279308 13783313 17493791 20922231 17718849 13794

provider(gendesc)
## [1] "UCSC"

seqinfo(gendesc)
## Seqinfo object with 7 sequences (1 circular) from ce2 genome:
##   seqnames seqlengths isCircular genome
##   chrI      15080483      FALSE   ce2
##   chrII      15279308      FALSE   ce2
##   chrIII     13783313      FALSE   ce2
##   chrIV      17493791      FALSE   ce2
##   chrV       20922231      FALSE   ce2
##   chrX       17718849      FALSE   ce2
##   chrM        13794        TRUE    ce2

bsgenomeName(gendesc)
## [1] "BSgenome.Celegans.UCSC.ce2"
```

3.2 SeqInfo class

```
## Note that all the arguments (except 'genome') must have the
## same length. 'genome' can be of length 1, whatever the lengths
## of the other arguments.
x <- Seqinfo(seqnames=c("chr1", "chr2", "chr3", "chrM"),
             seqlengths=c(100, 200, NA, 15),
             isCircular=c(NA, FALSE, FALSE, TRUE),
             genome="toy")

length(x)
## [1] 4

seqnames(x)
## [1] "chr1" "chr2" "chr3" "chrM"

names(x)
## [1] "chr1" "chr2" "chr3" "chrM"

seqlevels(x)
## [1] "chr1" "chr2" "chr3" "chrM"

seqlengths(x)
## chr1 chr2 chr3 chrM
## 100 200 NA 15
```



```

isCircular(x)

## chr1 chr2 chr3 chrM
##    NA FALSE FALSE  TRUE

genome(x)

## chr1 chr2 chr3 chrM
## "toy" "toy" "toy" "toy"

x[c("chrY", "chr3", "chr1")] # subset by names

## Seqinfo object with 3 sequences from 2 genomes (NA, toy):
##   seqnames seqlengths isCircular genome
##   chrY          NA          NA    <NA>
##   chr3          NA         FALSE    toy
##   chr1          100          NA     toy

## Rename, drop, add and/or reorder the sequence levels:
xx <- x
seqlevels(xx) <- sub("chr", "ch", seqlevels(xx)) # rename
xx

## Seqinfo object with 4 sequences (1 circular) from toy genome:
##   seqnames seqlengths isCircular genome
##   ch1          100          NA     toy
##   ch2          200         FALSE    toy
##   ch3          NA         FALSE    toy
##   chM          15          TRUE     toy

seqlevels(xx) <- rev(seqlevels(xx)) # reorder
xx

## Seqinfo object with 4 sequences (1 circular) from toy genome:
##   seqnames seqlengths isCircular genome
##   chM          15          TRUE     toy
##   ch3          NA         FALSE    toy
##   ch2          200         FALSE    toy
##   ch1          100          NA     toy

seqlevels(xx) <- c("ch1", "ch2", "chY") # drop/add/reorder
xx

## Seqinfo object with 3 sequences from 2 genomes (toy, NA):
##   seqnames seqlengths isCircular genome
##   ch1          100          NA     toy
##   ch2          200         FALSE    toy
##   chY          NA          NA    <NA>

seqlevels(xx) <- c(chY="Y", ch1="1", "22") # rename/reorder/drop/add
xx

## Seqinfo object with 3 sequences from 2 genomes (NA, toy):
##   seqnames seqlengths isCircular genome
##   Y          NA          NA    <NA>
##   1          100          NA     toy
##   22         NA          NA    <NA>

y <- Seqinfo(seqnames=c("chr3", "chr4", "chrM"),
              seqlengths=c(300, NA, 15))

```

```

y
## Seqinfo object with 3 sequences from an unspecified genome:
##   seqnames seqlengths isCircular genome
##   chr3      300        NA    <NA>
##   chr4      NA         NA    <NA>
##   chrM      15         NA    <NA>

merge(x, y) # rows for chr3 and chrM are merged

## Warning in .Seqinfo.mergexy(x, y): Each of the 2 combined objects has sequence levels not in
the other:
## - in 'x': chr1, chr2
## - in 'y': chr4
## Make sure to always combine/compare objects based on the same reference
## genome (use suppressWarnings() to suppress this warning).

## Seqinfo object with 5 sequences (1 circular) from 2 genomes (toy, NA):
##   seqnames seqlengths isCircular genome
##   chr1      100        NA    toy
##   chr2      200       FALSE    toy
##   chr3      300       FALSE    toy
##   chrM      15        TRUE    toy
##   chr4      NA         NA    <NA>

suppressWarnings(merge(x, y))

## Seqinfo object with 5 sequences (1 circular) from 2 genomes (toy, NA):
##   seqnames seqlengths isCircular genome
##   chr1      100        NA    toy
##   chr2      200       FALSE    toy
##   chr3      300       FALSE    toy
##   chrM      15        TRUE    toy
##   chr4      NA         NA    <NA>

## Note that, strictly speaking, merging 2 Seqinfo objects is not
## a commutative operation, i.e., in general 'z1 <- merge(x, y)'
## is not identical to 'z2 <- merge(y, x)'. However 'z1' and 'z2'
## are guaranteed to contain the same information (i.e. the same
## rows, but typically not in the same order):
suppressWarnings(merge(y, x))

## Seqinfo object with 5 sequences (1 circular) from 2 genomes (toy, NA):
##   seqnames seqlengths isCircular genome
##   chr3      300       FALSE    toy
##   chr4      NA         NA    <NA>
##   chrM      15        TRUE    toy
##   chr1      100        NA    toy
##   chr2      200       FALSE    toy

## This contradicts what 'x' says about circularity of chr3 and chrM:
isCircular(y)[c("chr3", "chrM")] <- c(TRUE, FALSE)
y

## Seqinfo object with 3 sequences (1 circular) from an unspecified genome:
##   seqnames seqlengths isCircular genome
##   chr3      300        TRUE    <NA>
##   chr4      NA         NA    <NA>
##   chrM      15        FALSE    <NA>

```

```
if (interactive()) {
  merge(x, y) # raises an error
}
```

4 Examples

4.1 converting seqlevel styles (eg:UCSC to NCBI)

A quick example using *Drosophila Melanogaster*. The txdb object contains seqlevels in UCSC style, we want to convert them to NCBI

```
txdb <- TxDb.Dmelanogaster.UCSC.dm3.ensGene
seqlevels(txdb)

## [1] "chr2L"      "chr2R"      "chr3L"      "chr3R"      "chr4"      "chrX"      "chrU"
## [8] "chrM"      "chr2LHet"   "chr2RHet"   "chr3LHet"   "chr3RHet"   "chrXHet"   "chrYHet"
## [15] "chrUextra"
```

```
genomeStyles("Drosophila melanogaster")

##      circular  sex  auto  NCBI      UCSC      Ensembl
## 1      FALSE FALSE  TRUE   2L     chr2L      2L
## 2      FALSE FALSE  TRUE   2R     chr2R      2R
## 3      FALSE FALSE  TRUE   3L     chr3L      3L
## 4      FALSE FALSE  TRUE   3R     chr3R      3R
## 5      FALSE FALSE  TRUE    4     chr4       4
## 6      FALSE  TRUE FALSE    X     chrX       X
## 7       TRUE FALSE FALSE   MT     chrM dmel_mitochondrion_genome
## 8      FALSE FALSE FALSE 2LHet  chr2LHet    2LHet
## 9      FALSE FALSE FALSE 2Rhet  chr2RHet    2RHet
## 10     FALSE FALSE FALSE 3LHet  chr3LHet    3LHet
## 11     FALSE FALSE FALSE 3Rhet  chr3RHet    3RHet
## 12     FALSE FALSE FALSE Xhet   chrXHet     XHet
## 13     FALSE FALSE FALSE Yhet   chrYHet     YHet
## 14     FALSE FALSE FALSE  Un     chrU       U
## 15     FALSE FALSE FALSE <NA> chrUextra  Uextra
```

```
mapSeqlevels(seqlevels(txdb), "NCBI")

##      chr2L      chr2R      chr3L      chr3R      chr4      chrX      chrU      chrM      chr2LHet
##      "2L"      "2R"      "3L"      "3R"      "4"      "X"      "Un"      "MT"      "2LHet"
## chr2RHet chr3LHet chr3RHet chrXHet chrYHet chrUextra
## "2Rhet"  "3LHet"  "3Rhet"  "Xhet"  "Yhet"      NA
```

4.2 converting styles and removing unwanted seqlevels

Suppose we read in a Bam file or a BED file and the resulting GRanges have a lot of seqlevels which are not required by your analysis or you want to rename the seqlevels from the current style to your own style (eg:UCSC to NCBI), we can use the functionality provided by *GenomeInfoDb* to do that.

Let us say that we have extracted the seqlevels of the Seqinfo object(say GRanges from a BED file) in a variable called "sequence".

```
sequence <- seqlevels(x)

## sequence is in UCSC format and we want NCBI style
newStyle <- mapSeqlevels(sequence,"NCBI")
newStyle <- newStyle[complete.cases(newStyle)] # removing NA cases.

## rename the seqlevels
x <- renameSeqlevels(x,newStyle)

## keep only the seqlevels you want (say autosomes)
auto <- extractSeqlevelsByGroup(species="Homo sapiens", style="NCBI",
                               group="auto")
x <- keepSeqlevels(x,auto)
```

5 Session Information

Here is the output of `sessionInfo` on the system on which this document was compiled:

```
toLatex(sessionInfo())
```

- R version 3.3.2 (2016-10-31), x86_64-w64-mingw32
- Locale: LC_COLLATE=C, LC_CTYPE=English_United States.1252, LC_MONETARY=English_United States.1252, LC_NUMERIC=C, LC_TIME=English_United States.1252
- Base packages: base, datasets, grDevices, graphics, methods, parallel, stats, stats4, utils
- Other packages: AnnotationDbi 1.36.2, BSgenome 1.42.0, BSgenome.Celegans.UCSC.ce2 1.4.0, Biobase 2.34.0, BiocGenerics 0.20.0, Biostrings 2.42.1, GenomeInfoDb 1.10.3, GenomicFeatures 1.26.2, GenomicRanges 1.26.2, IRanges 2.8.1, S4Vectors 0.12.1, TxDb.Dmelanogaster.UCSC.dm3.ensGene 3.2.2, XVector 0.14.0, rtracklayer 1.34.1
- Loaded via a namespace (and not attached): BiocParallel 1.8.1, BiocStyle 2.2.1, DBI 0.5-1, GenomicAlignments 1.10.0, Matrix 1.2-8, RCurl 1.95-4.8, RSQLite 1.1-2, Rcpp 0.12.9, Rsamtools 1.26.1, SummarizedExperiment 1.4.0, XML 3.98-1.5, biomaRt 2.30.0, bitops 1.0-6, digest 0.6.12, evaluate 0.10, grid 3.3.2, highr 0.6, knitr 1.15.1, lattice 0.20-34, magrittr 1.5, memoise 1.0.0, stringi 1.1.2, stringr 1.1.0, tools 3.3.2, zlibbioc 1.20.0