

An Introduction to *GenomeInfoDb*

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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"
## [3] "Canis_familiaris"          "Cyanidioschyzon_merolae"
## [5] "Drosophila_melanogaster"   "Gossypium_hirsutum"
## [7] "Homo_sapiens"              "Mus_musculus"
## [9] "Oryza_sativa"              "Populus_trichocarpa"
## [11] "Rattus_norvegicus"         "Saccharomyces_cerevisiae"
## [13] "Zea_mays"
```

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_gl000192_random", "4_ctg9_hap1"))  
seqlevelsInGroup(newchr, group="sex")
```

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```
## [1] "chrX" "chrY"

seqlevelsInGroup(newchr, group="auto")

## [1] "chr1"  "chr2"  "chr3"  "chr4"  "chr5"  "chr6"  "chr7"  "chr8"  "chr9"
## [10] "chr10" "chr11" "chr12" "chr13" "chr14" "chr15" "chr16" "chr17" "chr18"
## [19] "chr19" "chr20" "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.

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```
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      *
```

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```
## ... ...
## [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. Because these seqlevels are in use (i.e. have ranges on them), the ranges on these sequences need to be removed before the seqlevels can be dropped. We call this *pruning*. The `pruning.mode` argument controls how to prune gr. Unlike for list-like objects (e.g. `GRangesList`) for which pruning can be done in various ways, pruning a `GRanges` object is straightforward and achieved by specifying `pruning.mode="coarse"`.

```
dropSeqlevels(gr, paste0("chr",23:35), pruning.mode="coarse")

## 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), pruning.mode="coarse")

## 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 sequence style of the object.

```

keepStandardChromosomes(gr, pruning.mode="coarse")

## 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",
                       pruning.mode="coarse")

## 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 Seqinfo objects

```
## 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 are.
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
```

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```
## 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 .merge_two_Seqinfo_objects(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>
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>

## 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>
##   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>
##   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"
## [7] "chrU"        "chrM"        "chr2LHet"    "chr2RHet"    "chr3LHet"    "chr3RHet"
## [13] "chrXHet"    "chrYHet"    "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 FALSE  TRUE FALSE   Y     chrY            Y
## 8  TRUE FALSE FALSE  MT   chrM dmel_mitochondrion_genome
## 9 FALSE FALSE FALSE 2LHet chr2LHet          2LHet
## 10 FALSE FALSE FALSE 2RHET chr2RHET          2RHET
## 11 FALSE FALSE FALSE 3LHet chr3LHet          3LHet
## 12 FALSE FALSE FALSE 3RHET chr3RHET          3RHET
## 13 FALSE FALSE FALSE Xhet  chrXHet          XHet
## 14 FALSE FALSE FALSE Yhet  chrYHet          YHet
## 15 FALSE FALSE FALSE Un   chrU             U
## 16 FALSE FALSE FALSE <NA>  chrUextra        Uextra

mapSeqlevels(seqlevels(txdb), "NCBI")

##   chr2L     chr2R     chr3L     chr3R     chr4     chrX     chrU
##   "2L"     "2R"     "3L"     "3R"     "4"     "X"     "Un"
##   chrM     chr2LHet  chr2RHET  chr3LHet  chr3RHET  chrXHet  chrYHet
##   "MT"     "2LHet"   "2RHET"   "3LHet"   "3RHET"   "Xhet"   "Yhet"
##   chrUextra
##   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 4.4.2 (2024-10-31), x86_64-pc-linux-gnu
- Locale: LC_CTYPE=en_US.UTF-8, LC_NUMERIC=C, LC_TIME=en_GB, LC_COLLATE=C, LC_MONETARY=en_US.UTF-8, LC_MESSAGES=en_US.UTF-8, LC_PAPER=en_US.UTF-8, LC_NAME=C, LC_ADDRESS=C, LC_TELEPHONE=C, LC_MEASUREMENT=en_US.UTF-8, LC_IDENTIFICATION=C
- Time zone: America/New_York
- TZcode source: system (glibc)
- Running under: Ubuntu 24.04.1 LTS
- Matrix products: default
- BLAS: /home/biocbuild/bbs-3.20-bioc/R/lib/libRblas.so
- LAPACK: /usr/lib/x86_64-linux-gnu/lapack/liblapack.so.3.12.0
- Base packages: base, datasets, grDevices, graphics, methods, stats, stats4, utils
- Other packages: AnnotationDbi 1.68.0, Biobase 2.66.0, BiocGenerics 0.52.0, BiocStyle 2.34.0, GenomeInfoDb 1.42.3, GenomicFeatures 1.58.0, GenomicRanges 1.58.0, IRanges 2.40.1, S4Vectors 0.44.0, TxDb.Dmelanogaster.UCSC.dm3.ensGene 3.2.2
- Loaded via a namespace (and not attached): BiocIO 1.16.0, BiocManager 1.30.25, BiocParallel 1.40.0, Biostrings 2.74.1, DBI 1.2.3, DelayedArray 0.32.0, GenomeInfoDbData 1.2.13, GenomicAlignments 1.42.0, KEGGREST 1.46.0, Matrix 1.7-2, MatrixGenerics 1.18.1, R6 2.5.1, RCurl 1.98-1.16, RSQLite 2.3.9, Rsamtools 2.22.0, S4Arrays 1.6.0, SparseArray 1.6.1, SummarizedExperiment 1.36.0, UCSC.utils 1.2.0, XML 3.99-0.18, XVector 0.46.0, abind 1.4-8, bit 4.5.0.1, bit64 4.6.0-1, bitops 1.0-9, blob 1.2.4, bookdown 0.42, bslib 0.8.0, cachem 1.1.0, cli 3.6.3, codetools 0.2-20, compiler 4.4.2, crayon 1.5.3, curl 6.2.0, digest 0.6.37, evaluate 1.0.3, fastmap 1.2.0, grid 4.4.2, highr 0.11, htmltools 0.5.8.1, httr 1.4.7, jquerylib 0.1.4, jsonlite 1.8.9, knitr 1.49, lattice 0.22-6, lifecycle 1.0.4, matrixStats 1.5.0, memoise 2.0.1, parallel 4.4.2, pkgconfig 2.0.3, png 0.1-8, restfulr 0.0.15, rjson 0.2.23, rlang 1.1.5, rmarkdown 2.29, rtracklayer 1.66.0, sass 0.4.9, tools 4.4.2, vctrs 0.6.5, xfun 0.50, yaml 2.3.10, zlibbioc 1.52.0