

# An Introduction to *GenomeInfoDb*

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Modified: 26 June, 2020. Compiled: September 9, 2021

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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" "Homo_sapiens"
## [7] "Mus_musculus" "Oryza_sativa"
## [9] "Populus_trichocarpa" "Rattus_norvegicus"
## [11] "Saccharomyces_cerevisiae" "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")  
## [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.

```
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]   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
```

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. 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 *
```

	seqnames	ranges	strand
## [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
## [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
## [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
##   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
##   chrM      15        TRUE      toy
##   chr3      NA        FALSE     toy
##   chr2     200        FALSE     toy
##   chr1     100        NA        toy

seqlevels(xx) <- c("chr1", "chr2", "chrY") # drop/add/reorder
xx

## Seqinfo object with 3 sequences from 2 genomes (toy, NA):
##   seqnames seqlengths isCircular genome
##   chr1      100        NA        toy
##   chr2      200        FALSE     toy
##   chrY      NA        NA        <NA>

seqlevels(xx) <- c(chrY="Y", chr1="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"
## [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:USCS 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 4.1.1 (2021-08-10), 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
- Running under: Windows Server 2012 R2 x64 (build 9600)
- Matrix products: default
- Base packages: base, datasets, grDevices, graphics, methods, parallel, stats, stats4, utils
- Other packages: AnnotationDbi 1.54.1, Biobase 2.52.0, BiocGenerics 0.38.0, GenomeInfoDb 1.28.4, GenomicFeatures 1.44.2, GenomicRanges 1.44.0, IRanges 2.26.0, S4Vectors 0.30.0, TxDb.Dmelanogaster.UCSC.dm3.ensGene 3.2.2
- Loaded via a namespace (and not attached): BiocFileCache 2.0.0, BiocIO 1.2.0, BiocManager 1.30.16, BiocParallel 1.26.2, BiocStyle 2.20.2, Biostrings 2.60.2, DBI 1.1.1, DelayedArray 0.18.0, GenomeInfoDbData 1.2.6, GenomicAlignments 1.28.0, KEGGREST 1.32.0, Matrix 1.3-4, MatrixGenerics 1.4.3, R6 2.5.1, RCurl 1.98-1.4, RSQLite 2.2.8, Rcpp 1.0.7, Rsamtools 2.8.0, SummarizedExperiment 1.22.0, XML 3.99-0.7, XVector 0.32.0, assertthat 0.2.1, biomaRt 2.48.3, bit 4.0.4, bit64 4.0.5, bitops 1.0-7, blob 1.2.2, cachem 1.0.6, compiler 4.1.1, crayon 1.4.1, curl 4.3.2, dbplyr 2.1.1, digest 0.6.27, dplyr 1.0.7, ellipsis 0.3.2, evaluate 0.14, fansi 0.5.0, fastmap 1.1.0, filelock 1.0.2, generics 0.1.0, glue 1.4.2, grid 4.1.1, highr 0.9, hms 1.1.0, htmltools 0.5.2, httr 1.4.2, knitr 1.34, lattice 0.20-44, lifecycle 1.0.0, magrittr 2.0.1, matrixStats 0.60.1, memoise 2.0.0, pillar 1.6.2, pkgconfig 2.0.3, png 0.1-7, prettyunits 1.1.1, progress 1.2.2, purrr 0.3.4, rappdirs 0.3.3, restfulr 0.0.13, rjson 0.2.20, rlang 0.4.11, rmarkdown 2.10, rtracklayer 1.52.1, stringi 1.7.4, stringr 1.4.0, tibble 3.1.4, tidyselect 1.1.1, tools 4.1.1, utf8 1.2.2, vctrs 0.3.8, xfun 0.25, xml2 1.3.2, yaml 2.2.1, zlibbioc 1.38.0