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 exsiting organisms

2.1 genomeStyles

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

```
seqmap <- genomeStyles()</pre>
head(seqmap, n = 2)
## $Arabidopsis_thaliana
##
     circular auto
                       sex NCBI TAIR10
## 1
        FALSE TRUE FALSE
                              1
                                     1
                                     2
        FALSE TRUE FALSE
## 3
        FALSE TRUE FALSE
                              3
                                     3
        FALSE TRUE FALSE
```

```
## 5
       FALSE TRUE FALSE
                                   5
                          5
## 6
        TRUE FALSE FALSE
                                  Mt
## 7
       FALSE FALSE TRUE Pltd
                                  Pt.
##
## $Caenorhabditis_elegans
                                UCSC Ensembl
##
    circular auto sex NCBI
## 1
       FALSE TRUE FALSE
                          I
                                chrI
                                          Ι
## 2
       FALSE TRUE FALSE
                           II chrII
                                         TT
## 3
       FALSE TRUE FALSE III chrIII
                                         III
       FALSE TRUE FALSE
                          IV chrIV
                                         IV
## 4
       FALSE TRUE FALSE
                           V
                                          V
                                chrV
       FALSE FALSE TRUE
                                          χ
## 6
                           X
                                chrX
     TRUE TRUE FALSE
                         MT
                                chrM
                                       MtDNA
```

Oragnism's supported by GenomeInfoDb can be found by :

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)
                     sex NCBI UCSC
##
     circular auto
## 1
        FALSE TRUE FALSE
                            1 chr1
## 2
        FALSE TRUE FALSE
                            2 chr2
        FALSE TRUE FALSE
                            3 chr3
        FALSE TRUE FALSE
                            4 chr4
       FALSE TRUE FALSE
                          5 chr5
```

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 seglevelsStyle

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 seglevelsInGroup

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 prvoide 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" "chr10"

## [11] "chr11" "chr12" "chr13" "chr14" "chr15" "chr16" "chr17" "chr18" "chr19" "chr20"

## [21] "chr21" "chr22"

seqlevelsInGroup(newchr, group = "circular")

## [1] "chrX" "chrY"</pre>
```

if we have a vector conatining 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</pre>
```

2.6 orderSeglevels

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</pre>
```

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</pre>
```

2.8 mapSeglevels

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"
```

3 Examples

3.1 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
         FALSE FALSE
                                                                    ЗL
## 3
                      TRUE
                               3L
                                      chr3L
## 4
         FALSE FALSE TRUE
                               3R
                                      chr3R
                                                                    3R
         FALSE FALSE TRUE
                                                                     4
## 5
                                4
                                       chr4
## 6
         FALSE TRUE FALSE
                                Χ
                                       chrX
                                                                     Χ
## 7
         TRUE FALSE FALSE
                               MT
                                       chrM dmel_mitochondrion_genome
## 8
         FALSE FALSE FALSE 2LHet
                                  chr2LHet
                                                                 2LHet
## 9
         FALSE FALSE FALSE 2Rhet
                                  chr2RHet
                                                                 2RHet
         FALSE FALSE SLHet
## 10
                                   chr3LHet
                                                                 3LHet
## 11
         FALSE FALSE FALSE 3RHet
                                   chr3RHet
                                                                 3RHet
## 12
         FALSE FALSE FALSE
                            Xhet
                                    chrXHet
                                                                  XHet
         FALSE FALSE FALSE
                                    chrYHet
                                                                  YHet
## 13
                            Yhet
## 14
         FALSE FALSE FALSE
                               Un
                                       chrU
                                                                     U
## 15
         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 | | | | |

3.2 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 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)</pre>
```