An Introduction to the GenomicAlignments Package

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Contents

1	Introduction	1	
2	GAlignments: Genomic Alignments 2.1 Load a 'BAM' file into a GAlignments object 2.2 Simple accessor methods 2.3 More accessor methods	3	
3	<i>GAlignmentPairs</i> : Pairs of Genomic Alignments		
4	GAlignmentsList: Groups of Genomic Alignments	4	
5	Session Information	4	

1 Introduction

The *GenomicAlignments* package serves as the foundation for representing genomic alignments within the Bioconductor project. In the Bioconductor package hierarchy, it builds upon the *GenomicRanges* (infrastructure) package and provides support for many Bioconductor packages.

This package defines three classes: *GAlignments*, *GAlignmentPairs*, and *GAlignmentsList*), which are used to represent genomic alignments, pairs of genomic alignments, and groups of genomic alignments.

The GenomicAlignments package is available at bioconductor.org and can be downloaded via BiocManager::install:

> if (!require("BiocManager"))
+ install.packages("BiocManager")
> BiocManager::install("GenomicAlignments")

> library(GenomicAlignments)

2 GAlignments: Genomic Alignments

The *GAlignments* class which is a container for storing a set of genomic alignments. The class is intended to support alignments in general, not only those coming from a 'Binary Alignment Map' or 'BAM' files. Also alignments with gaps in the reference sequence (a.k.a. *gapped alignments*) are supported which, for example, makes the class suited for storing junction reads from an RNA-seq experiment.

More precisely, a *GAlignments* object is a vector-like object where each element describes an *alignment*, that is, how a given sequence (called *query* or *read*, typically short) aligns to a reference sequence (typically long).

As shown later in this document, a *GAlignments* object can be created from a 'BAM' file. In that case, each element in the resulting object will correspond to a record in the file. One important thing to note though is that not all

the information present in the BAM/SAM records is stored in the object. In particular, for now, we discard the query sequences (SEQ field), the query ids (QNAME field), the query qualities (QUAL), the mapping qualities (MAPQ) and any other information that is not needed in order to support the basic set of operations described in this document. This also means that multi-reads (i.e. reads with multiple hits in the reference) don't receive any special treatment i.e. the various SAM/BAM records corresponding to a multi-read will show up in the *GAlignments* object as if they were coming from different/unrelated queries. Also paired-end reads will be treated as single-end reads and the pairing information will be lost. This might change in the future.

2.1 Load a 'BAM' file into a GAlignments object

First we use the readGAlignments function from the *GenomicAlignments* package to load a toy 'BAM' file into a *GAlignments* object:

```
> library(GenomicAlignments)
> aln1 file <- system.file("extdata", "ex1.bam", package="Rsamtools")</pre>
> aln1 <- readGAlignments(aln1_file)</pre>
> aln1
GAlignments object with 3271 alignments and 0 metadata columns:
          seqnames strand
                            cigar
                                            qwidth
                                                        start
                                                                     end
             <Rle> <Rle> <character> <integer> <integer> <integer>
     [1]
              seq1
                      +
                                    36M
                                                36
                                                            1
                                                                       36
                                    35M
                                                35
                                                            3
                                                                       37
     [2]
              seq1
                         +
     [3]
              seq1
                         ^+
                                    35M
                                                35
                                                            5
                                                                      39
     [4]
                         +
                                    36M
                                                36
                                                            6
                                                                      41
              seq1
                                                            9
     [5]
              seq1
                         ^+
                                    35M
                                                35
                                                                      43
     . . .
               . . .
                       . . .
                                    . . .
                                               . . .
                                                          . . .
                                                                      . . .
  [3267]
                         ^+
                                    35M
                                                35
                                                         1524
                                                                    1558
              seq2
  [3268]
              seq2
                         +
                                    35M
                                                35
                                                         1524
                                                                    1558
  [3269]
              seq2
                         _
                                    35M
                                                35
                                                         1528
                                                                    1562
                         _
                                    35M
                                                35
                                                                    1566
  [3270]
              seq2
                                                         1532
                         _
                                    35M
                                                35
                                                         1533
                                                                    1567
  [3271]
              seq2
              width
                        njunc
          <integer> <integer>
                 36
                             0
     [1]
     [2]
                 35
                             0
                 35
                             0
     [3]
     [4]
                 36
                             0
                 35
                             0
     [5]
     . . .
                . . .
                           . . .
  [3267]
                 35
                             0
  [3268]
                 35
                             0
                 35
                             0
  [3269]
                 35
                             0
  [3270]
                 35
  [3271]
                             0
  _____
  seqinfo: 2 sequences from an unspecified genome
```

> length(aln1)

[1] 3271

3271 'BAM' records were loaded into the object.

Note that readGAlignments would have discarded any 'BAM' record describing an unaligned query (see description of the <flag> field in the SAM Format Specification ¹ for more information). The reader interested in tracking down these events can always use the scanBam function but this goes beyond the scope of this document.

2.2 Simple accessor methods

There is one accessor per field displayed by the show method and it has the same name as the field. All of them return a vector or factor of the same length as the object:

```
> head(seqnames(aln1))
factor-Rle of length 6 with 1 run
 Lengths:
              6
 Values : seq1
Levels(2): seq1 seq2
> seqlevels(aln1)
[1] "seq1" "seq2"
> head(strand(aln1))
factor-Rle of length 6 with 1 run
 Lengths: 6
 Values : +
Levels(3): + - *
> head(cigar(aln1))
[1] "36M" "35M" "35M" "36M" "35M" "35M"
> head(qwidth(aln1))
[1] 36 35 35 36 35 35
> head(start(aln1))
[1] 1 3 5 6 9 13
> head(end(aln1))
[1] 36 37 39 41 43 47
> head(width(aln1))
[1] 36 35 35 36 35 35
> head(njunc(aln1))
[1] 0 0 0 0 0 0
```

2.3 More accessor methods

[coming soon...]

¹http://samtools.sourceforge.net/SAM1.pdf

3 GAlignmentPairs: Pairs of Genomic Alignments

[coming soon...]

4 GAlignmentsList: Groups of Genomic Alignments

[coming soon...]

5 Session Information

All of the output in this vignette was produced under the following conditions:

```
> sessionInfo()
R Under development (unstable) (2024-10-21 r87258)
Platform: x86 64-pc-linux-gnu
Running under: Ubuntu 24.04.1 LTS
Matrix products: default
BLAS: /home/biocbuild/bbs-3.21-bioc/R/lib/libRblas.so
LAPACK: /usr/lib/x86_64-linux-gnu/lapack/liblapack.so.3.12.0
locale:
[1] LC_CTYPE=en_US.UTF-8 LC_NUMERIC=C
                              LC_COLLATE=C
 [3] LC_TIME=en_GB
 [5] LC_MONETARY=en_US.UTF-8 LC_MESSAGES=en_US.UTF-8
[7] LC_PAPER=en_US.UTF-8 LC_NAME=C
 [9] LC_ADDRESS=C
                              LC_TELEPHONE=C
[11] LC_MEASUREMENT=en_US.UTF-8 LC_IDENTIFICATION=C
time zone: America/New_York
tzcode source: system (glibc)
attached base packages:
[1] stats4 stats graphics grDevices utils
                                                   datasets
[7] methods base
other attached packages:
 [1] pasillaBamSubset_0.43.0
 [2] GenomicAlignments_1.43.0
 [3] SummarizedExperiment_1.37.0
 [4] Biobase_2.67.0
 [5] MatrixGenerics_1.19.0
 [6] matrixStats_1.4.1
 [7] Rsamtools_2.23.0
 [8] Biostrings_2.75.0
 [9] XVector_0.47.0
[10] GenomicRanges_1.59.0
[11] GenomeInfoDb_1.43.0
[12] IRanges_2.41.0
[13] S4Vectors_0.45.0
[14] BiocGenerics_0.53.0
[15] RNAseqData.HNRNPC.bam.chr14_0.43.0
[16] BiocStyle_2.35.0
```

loaded via a namespace (and not attached):				
	-			
[1]	Matrix_1.7-1	jsonlite_1.8.9		
[3]	compiler_4.5.0	BiocManager_1.30.25		
[5]	crayon_1.5.3	bitops_1.0-9		
[7]	parallel_4.5.0	jquerylib_0.1.4		
[9]	BiocParallel_1.41.0	yaml_2.3.10		
[11]	fastmap_1.2.0	lattice_0.22-6		
[13]	R6_2.5.1	S4Arrays_1.7.0		
[15]	knitr_1.48	DelayedArray_0.33.0		
[17]	bookdown_0.41	GenomeInfoDbData_1.2.13		
[19]	bslib_0.8.0	rlang_1.1.4		
[21]	cachem_1.1.0	xfun_0.48		
[23]	sass_0.4.9	SparseArray_1.7.0		
[25]	cli_3.6.3	zlibbioc_1.53.0		
[27]	grid_4.5.0	digest_0.6.37		
[29]	lifecycle_1.0.4	evaluate_1.0.1		
[31]	codetools_0.2-20	abind_1.4-8		
[33]	rmarkdown_2.28	httr_1.4.7		
[35]	tools_4.5.0	htmltools_0.5.8.1		
[37]	UCSC.utils_1.3.0			