Accessing Genome annotations from the UCSC Genome Browser

Marc Carlson

December 6, 2010

1 Introduction

The *rtacklayer* package provides functions and methods that can be used to get the data tables behind the UCSC tracks and import them as data.frames. This vignette will explore some of these and document the capabilities with specific examples.

1.1 Retrieving Exon Boundary information

In general, when you want to get some data from UCSC, you will want to 1st make a session. The most common thing is that you will want a session with the UCSC Genome Browser, so this is the default behavior.

```
> library(rtracklayer)
```

```
> session <- browserSession()</pre>
```

Once you have done this, you will need to choose which genome you want to work on. To do that, you should use the ucscGenomes function to list all the available genomes and then choose one as follows.

```
> head(ucscGenomes())
```

	db	$\operatorname{organism}$		date			name
1	hg19	Human	Feb.	2009	Genome	Reference	Consortium GRCh37
2	hg18	Human	Mar.	2006			NCBI Build 36.1
3	hg17	Human	May	2004			NCBI Build 35
4	hg16	Human	Jul.	2003			NCBI Build 34
5	felCat4	Cat	Dec.	2008			NHGRI catChrV17e
6	felCat3	Cat	Mar.	2006		Broad I	nstitute Release 3

Then you can set the value of the chosen genome for your session using the **genome** command. The following command sets it to be human build hg18.

```
> genome(session) <- "hg18"</pre>
```

To search for tracks/tables are available you can use the trackNames method like this:

```
> head(trackNames(session))
```

Base Position Chromosome Band STS Markers FISH Clones Recomb Rate "ruler" "cytoBand" "stsMap" "fishClones" "recombRate" Map Contigs "ctgPos"

Finally, you can retrieve the data from UCSC by using the ucscTable-Query command. In this case we just want to get the whole table so we will leave out the option of passing in the segment of the genome we would want to retrieve it for. The following example will create a query to retrieve the entire table/track for the refGene track from mouse.

```
> query <- ucscTableQuery(session, "refGene")</pre>
```

Then we can use the getTable method to return the data in the query.

```
> head(getTable(query))
```

1.2 Some other Resources

Several kinds of data are available for access. Here are some tracks from human, that I expect are likely to be popular:

CPG Islands: "cpgIslandExt"

Access to genes known to be associated with disease: "gad", "omimGene" Nucleosome Occupancy: "uwNucOcc" (this one is causing trouble) Genomic Segmental Duplications: "genomicSuperDups" Conserved TFBS: "tfbsConsSites"

1.3 Restricting annotations to a Genomic Region

Sometimes you may also want to restrict the amount of data you retrieve. In these cases you can pass a GenomicRanges object in to the ucscTableSession so that it will limit the values returned to only the region of interest. This can be especially true when looking at data that occurs in a lot of places in the genome such as SNPs. Below is an example that will return the SNPs on a particular region of Chromosome 12.

```
> guery <- ucscTableQuery(session, "snp130",</pre>
+
                            GenomicRanges(57795963, 57815592, "chr12"))
> head(getTable(query))
                                         name score strand refNCBI refUCSC
   bin chrom chromStart chromEnd
1 1025 chr12
                57796046 57796047 rs12822426
                                                                   С
                                                                           С
                                                   0
                                                          +
2 1025 chr12
                                                                   G
                                                                           G
                57796211 57796212 rs1356171
                                                   0
                                                          _
3 1025 chr12
                57796920 57796920 rs34250032
                                                   0
                                                          +
                                                                   _
                                                                           _
                                                                   С
                                                                           С
4 1025 chr12
                57797200 57797201 rs2120529
                                                   0
                                                          _
5 1025 chr12
                57797278 57797279 rs2120528
                                                   0
                                                                   Т
                                                                           Т
6 1025 chr12
                57797524 57797525 rs12831695
                                                   0
                                                                   Т
                                                                           Т
                                                          +
  observed molType
                        class
1
       C/T genomic
                       single
2
       C/T genomic
                       single
3
       -/C genomic insertion
4
       A/G genomic
                       single
5
       A/T genomic
                       single
6
       G/T genomic
                       single
                                                                          avHet
                                                                 valid
                                   by-cluster, by-frequency, by-hapmap 0.087777
1
2
                                                               unknown 0.000000
3
                                                               unknown 0.000000
4 by-cluster, by-frequency, by-2hit-2allele, by-hapmap, by-1000genomes 0.175690
5 by-cluster, by-frequency, by-2hit-2allele, by-hapmap, by-1000genomes 0.340527
6
                                                               unknown 0.000000
   avHetSE
               func locType weight
1 0.190220 unknown
                      exact
                                  1
2 0.000000 unknown
                                  1
                      exact
3 0.000000 unknown between
                                  1
4 0.238701 unknown
                                  1
                      exact
5 0.233034 unknown
                                  1
                      exact
6 0.000000 unknown
                      exact
                                  1
```

1.4 Even More Resources

Here are some additional types of information that are expected to be popular:

Mapped Ests: "est" Recombination Rates: "recombRate" Microsattelites: "microsat" Comparative genomics information: "chainBosTau4" (eg. compare w/bovines)

2 Session Information

The version number of R and packages loaded for generating the vignette were:

```
R version 2.12.0 Patched (2010-11-28 r53696)
Platform: x86_64-unknown-linux-gnu (64-bit)
```

```
locale:
```

```
[1] LC_CTYPE=en_US.UTF-8
                                LC_NUMERIC=C
 [3] LC_TIME=en_US.UTF-8
                                LC_COLLATE=C
 [5] LC_MONETARY=C
                                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
attached base packages:
[1] stats
              graphics grDevices utils
                                            datasets methods
                                                                 base
other attached packages:
[1] rtracklayer_1.10.5
                          RCurl_1.5-0
                                                 bitops_1.0-4.1
[4] GenomicFeatures_1.2.3 GenomicRanges_1.2.1
                                                 IRanges_1.8.5
loaded via a namespace (and not attached):
[1] BSgenome_1.18.2
                      Biobase_2.10.0
                                        Biostrings_2.18.2 DBI_0.2-5
[5] RSQLite_0.9-4
                      XML_3.2-0
                                        biomaRt_2.6.0
                                                           tools_2.12.0
```