The GenomeGraphs user's guide

Steffen Durinck*and James Bullard†

May 2, 2019

Contents

1 Introduction

Genomic data analyses can benifit from integrated visualization of the genomic information. The GenomeGraphs package uses the biomaRt package to do live queries to Ensembl and translates e.g. gene/transcript structures to viewports of the grid graphics package, resulting in genomic information plotted together with your data. Possible genomics datasets that can be plotted are: Array CGH data, gene expression data and sequencing data.

*sdurinck@gmail.com
†bullard@stat.berkeley.edu

```
> library(GenomeGraphs)
```

2 Creating a Ensembl annotation graphic

To create an Ensembl annotation graphic, you need to decide what you want to plot. Genes and transcripts can be plotted individually using the `Gene` and `Transcript` objects respectively. Or one can plot a gene region the forward strand or reverse strand only or both. In this section we will cover these different graphics.

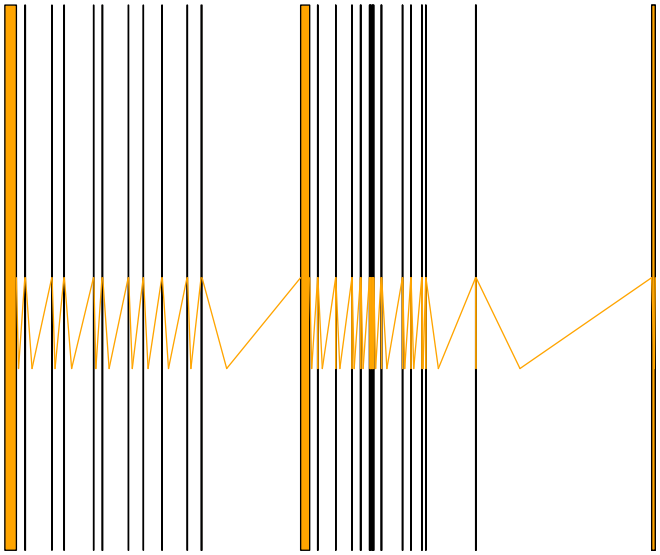
2.1 Plotting a Gene

If one wants to plot annotation information from Ensembl then you need to connect to the Ensembl BioMart database using the useMart function of the biomaRt package.

```
> mart <- useMart("ensembl", dataset="hsapiens_gene_ensembl")
```

Next we can retrieve the gene structure of the gene of interest.

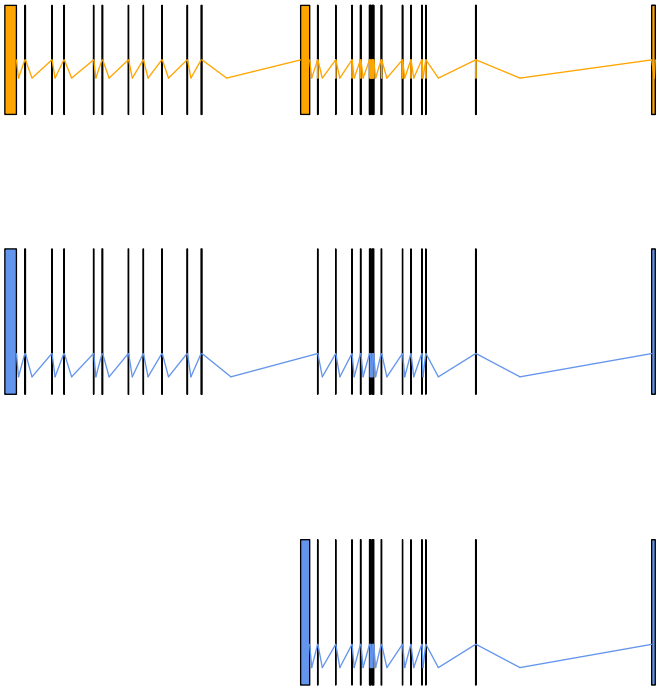
```
> gene <- makeGene(id = "ENSG00000095203", type="ensembl_gene_id", biomart = mart)
> gdPlot(gene)
```



2.2 Adding alternative transcripts

To add alternative transcripts you first have to create a `Transcript` object. Note that the order of the objects in the list determines the order in the plot.

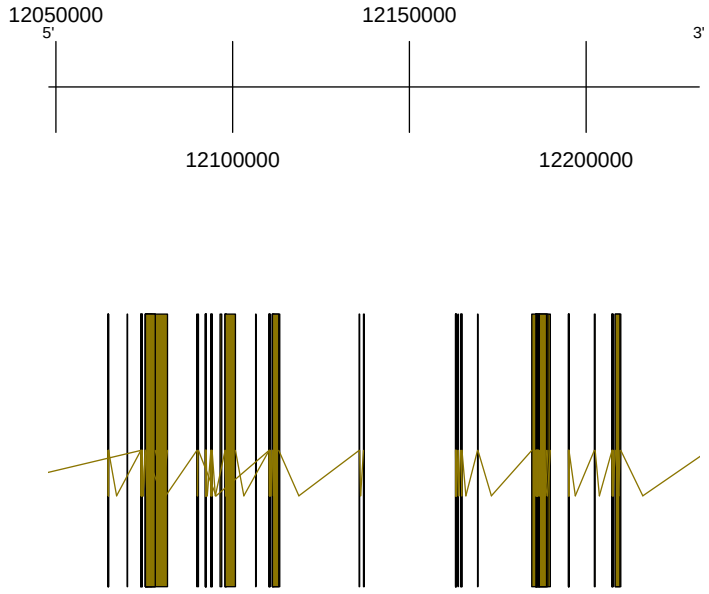
```
> transcript <- makeTranscript(id = "ENSG00000095203", type="ensembl_gene_id", biomar
> gdPlot(list(gene, transcript))
```



2.3 Plotting a gene region

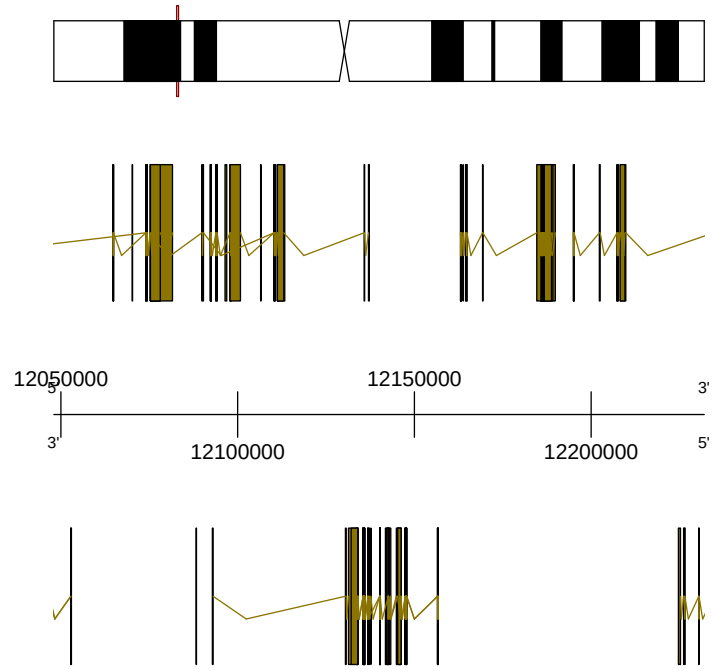
If you're interested in not just plotting one gene but a whole gene region the you should create a `GeneRegion` object. Note that a `GeneRegion` object is strand specific. In the example below we will retrieve the genes on the forward (+) strand only and add a genomic axis as well to give us the base positions.

```
> plusStrand <- makeGeneRegion(chromosome = 19, start = 12050000, end = 12230000, str
> genomeAxis <- makeGenomeAxis(add53 = TRUE)
> gdPlot(list(genomeAxis, plusStrand))
```



Let's now add the genes on the negative strand as well and an ideogram of chromosome 17, highlighting the region we are looking at.

```
> minStrand <- makeGeneRegion( chromosome = 19, start = 12050000, end = 12230000, str
> ideogram <- makeIdeogram(chromosome = 19)
> genomeAxis <- makeGenomeAxis(add53=TRUE, add35=TRUE)
> gdPlot(list(ideogram, plusStrand, genomeAxis, minStrand))
```



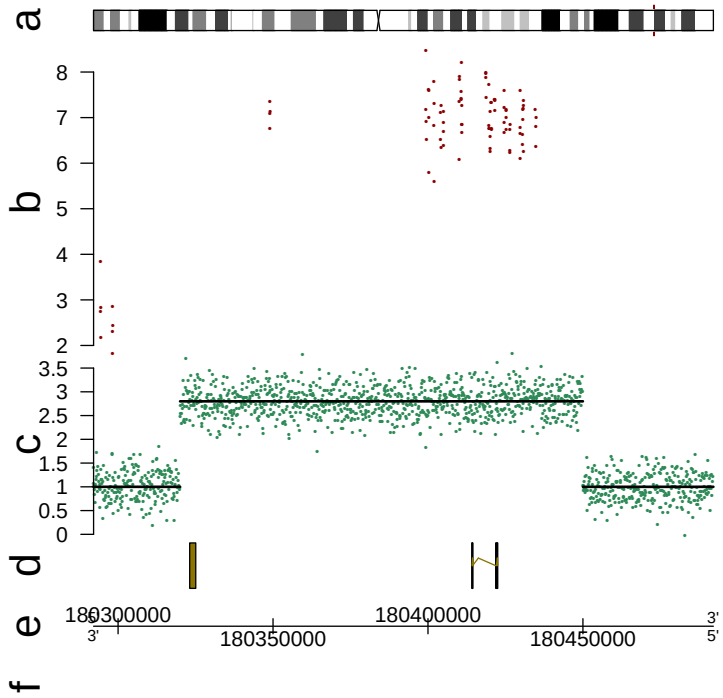
3 Adding Array data to the plot

3.1 Array CGH and gene expression array data

The `Generic Array` object enables plotting of expression and CGH array data together with segments if available. The array intensity data should be given as a matrix, with in the rows t he different probes and in the columns the different samples. For each probe the start location should be given using the probeStart argument. This should be a one column matrix. Lets load some dummy data.

```
> data("exampleData", package="GenomeGraphs")
> minbase <- 180292097
> maxbase <- 180492096
> genesplus <- makeGeneRegion(start = minbase, end = maxbase,
+                             strand = "+", chromosome = "3", biomart=mart)
> genesmin <- makeGeneRegion(start = minbase, end = maxbase,
+                            strand = "-", chromosome = "3", biomart=mart)
```

```
> seg <- makeSegmentation(segStart[[1]], segEnd[[1]], segments[[1]],
+                         dp = DisplayPars(color = "black", lwd=2,lty = "solid"))
> cop <- makeGenericArray(intensity  = cn, probeStart = probestart,
+                         trackOverlay =  seg, dp = DisplayPars(size=3, color = "seag
> ideog <- makeIdeogram(chromosome = 3)
> expres <- makeGenericArray(intensity = intensity, probeStart = exonProbePos,
+                            dp = DisplayPars(color="darkred", type="point"))
> genomeAxis <- makeGenomeAxis(add53 = TRUE, add35=TRUE)
> gdPlot(list(a=ideog,b=expres,c=cop,d=genesplus,e=genomeAxis,f=genesmin),
+        minBase = minbase, maxBase =maxbase, labelCex = 2)
```

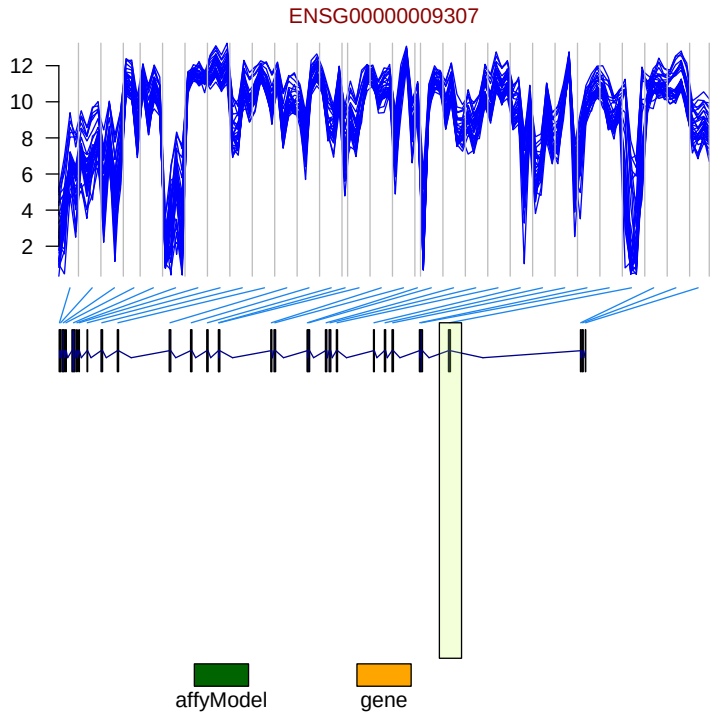


3.2 Exon array data

The example below plots probe level exon array data and is usefull in relating alternative splicing with known transcript structures.

```
> data("unrData", package="GenomeGraphs")
> title <- makeTitle(text ="ENSG00000009307", color = "darkred")
```

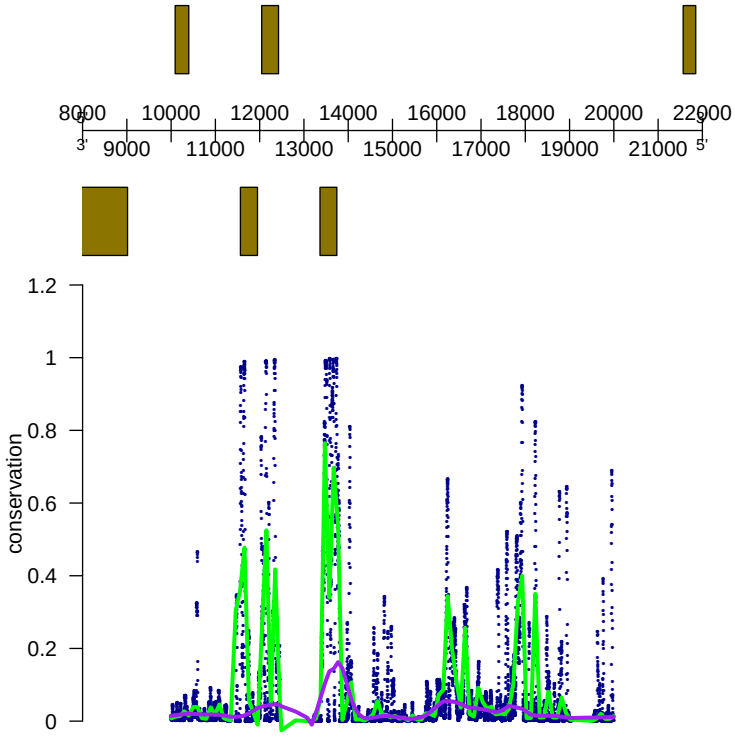
```
> exon <- makeExonArray(intensity = unrData, probeStart = unrPositions[,3],
+             probeEnd=unrPositions[,4], probeId = as.character(unrPositions[,1]),
+             nProbes = unrNProbes, dp = DisplayPars(color = "blue", mapColor = "dodg
+             displayProbesets=FALSE)
> affyModel.model <- makeGeneModel(start = unrPositions[,3], end = unrPositions[,4])
> affyModel <- makeAnnotationTrack(start = unrPositions[,3], end = unrPositions[,4],
+                                  feature = "gene_model", group = "ENSG00000009307",
+                                  dp = DisplayPars(gene_model = "darkblue"))
> gene <- makeGene(id = "ENSG00000009307", biomart = mart)
> transcript <- makeTranscript( id ="ENSG00000009307" , biomart = mart)
> legend <- makeLegend(c("affyModel","gene"), fill = c("darkgreen","orange"))
> rOverlay <- makeRectangleOverlay(start = 115085100, end = 115086500, region = c(3,5
+                                  dp = DisplayPars(alpha = .2, fill = "olivedrab1"))
> gdPlot(list(title, exon, affyModel, gene, transcript, legend),
+        minBase = 115061061, maxBase=115102147, overlay = rOverlay)
```



3.3 Plotting Conservation Data

The UCSC genome browser offers downloadable conservation data for a variety of species. Here we show how you can plot that conservation data along with annotation.

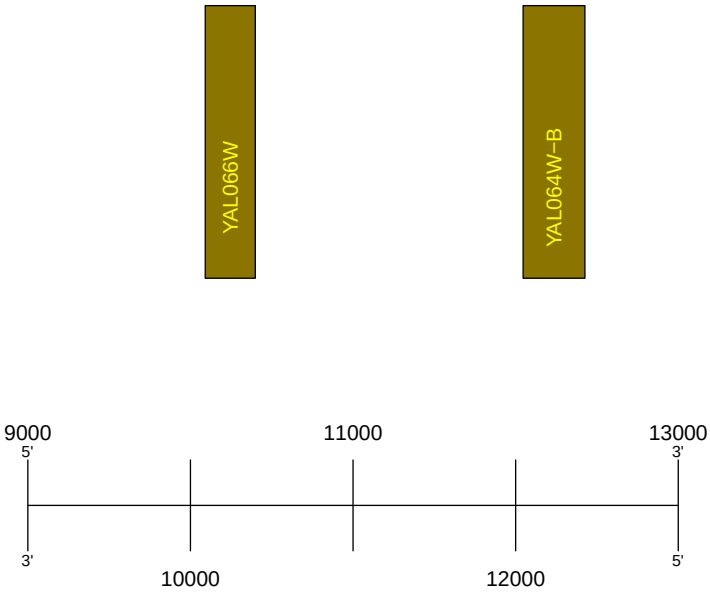
```
> yeastMart <- useMart("ensembl", dataset = "scerevisiae_gene_ensembl")
> minB <- 10000
> maxB <- 20000
> chrRoman <- as.character(as.roman(1))
> grP <- makeGeneRegion(start = minB, end = maxB, strand = "+",
+                       chromosome = chrRoman, biomart = yeastMart)
> grM <- makeGeneRegion(start = minB, end = maxB, strand = "-",
+                       chromosome = chrRoman, biomart = yeastMart)
> gaxis <- makeGenomeAxis(add53 = TRUE, add35 = TRUE)
> conserv <- yeastCons1[yeastCons1[,1] > minB & yeastCons1[,1] < maxB, ]
> s1 <- makeSmoothing(x = lowess(conserv[,1], conserv[,2], f = .01)$x,
+                     y = lowess(conserv[,1], conserv[,2], f = .01)$y,
+                     dp = DisplayPars(lwd = 3, color = "green"))
> s2 <- makeSmoothing(x = lowess(conserv[,1], conserv[,2], f = .1)$x,
+                     y = lowess(conserv[,1], conserv[,2], f = .1)$y,
+                     dp = DisplayPars(lwd = 3, color = "purple"))
> consTrack <- makeBaseTrack(base = conserv[, 1], value = conserv[,2],
+                            dp = DisplayPars(lwd=.2, ylim = c(0, 1.25),
+                            color = "darkblue"), trackOverlay = list(s1, s2))
> gdPlot(list(grP, gaxis, grM, "conservation" = consTrack))
```



4 Odds and Ends

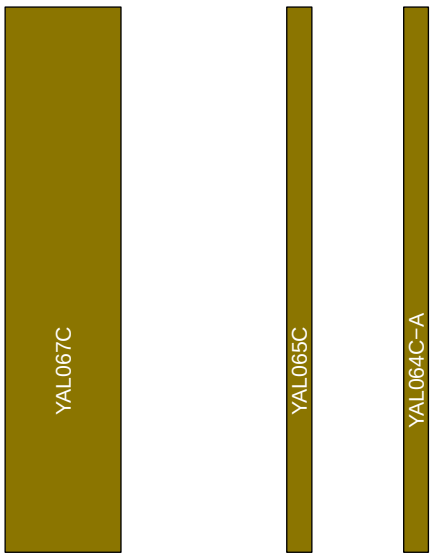
In addition to plotting the genes we can enable the plotting of names of genes.

```
> plotGeneRegion <- function(chr = 1, minB = 9000, maxB = 13000, rot = 0, col = "gree
+   chrRoman <- as.character(as.roman(1:17)[chr])
+   grP <- makeGeneRegion(start = minB, end = maxB,
+                         strand = "+", chromosome = chrRoman, biomart = yeastMart,
+                         dp = DisplayPars(plotId = TRUE, idRotation = rot,
+                         idColor = col))
+   gaxis <- makeGenomeAxis( add53 = TRUE, add35 = TRUE, littleTicks = FALSE)
+   gdPlot(list(grP, gaxis), minBase = minB, maxBase = maxB)
+ }
> plotGeneRegion(col = "yellow", rot=90)
```



Finally, if you are interested in seeing how things look you can just plot the object without the list, or without the *minBase*, *maxBase* arguments.

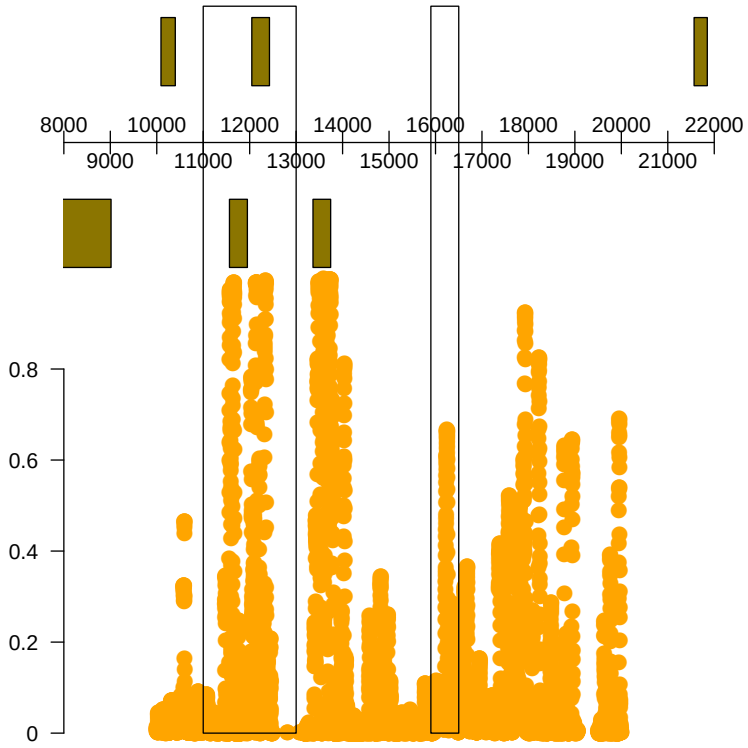
```
> gdPlot(makeGeneRegion(start = 9000, end = 15000, biomart = yeastMart,
+                       strand = "-", chromosome = "I",
+                       dp = DisplayPars(plotId=TRUE)))
```



4.1 Overlays

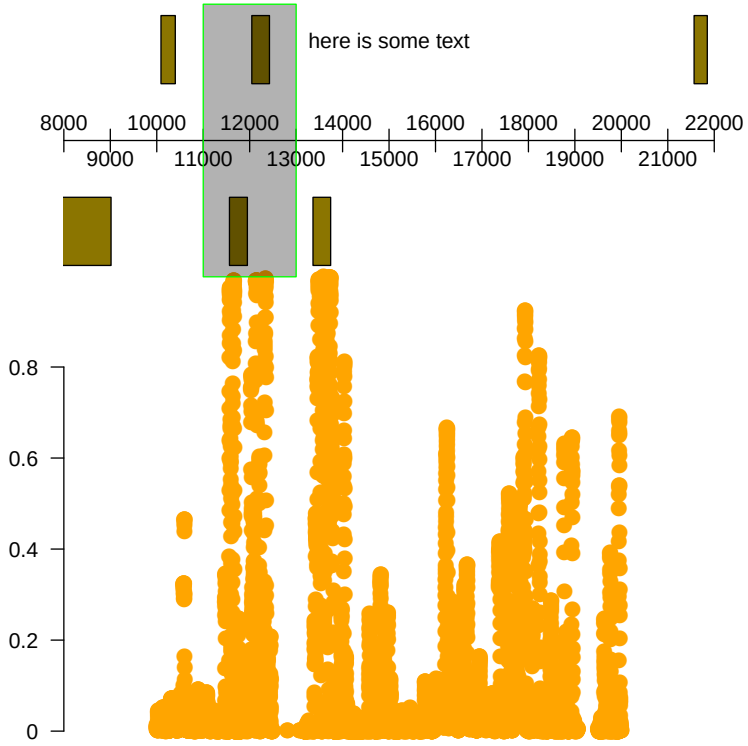
`Overlays` can be used to annotate different regions of the plot. Currently, we can draw boxes and write text on the plot.

```
> ga <- makeGenomeAxis()
> grF <- makeGeneRegion(start = 10000, end = 20000, chromosome = "I", strand = "+", b
> grR <- makeGeneRegion(start = 10000, end = 20000, chromosome = "I", strand = "-", b
> bt <- makeBaseTrack(base = yeastCons1[,1], value = yeastCons1[,2])
> hr1 <- makeRectangleOverlay(start = 11000, end = 13000)
> hr2 <- makeRectangleOverlay(start = 15900, end = 16500)
> gdPlot(list(grF, ga, grR, bt), overlays = list(hr1, hr2))
```



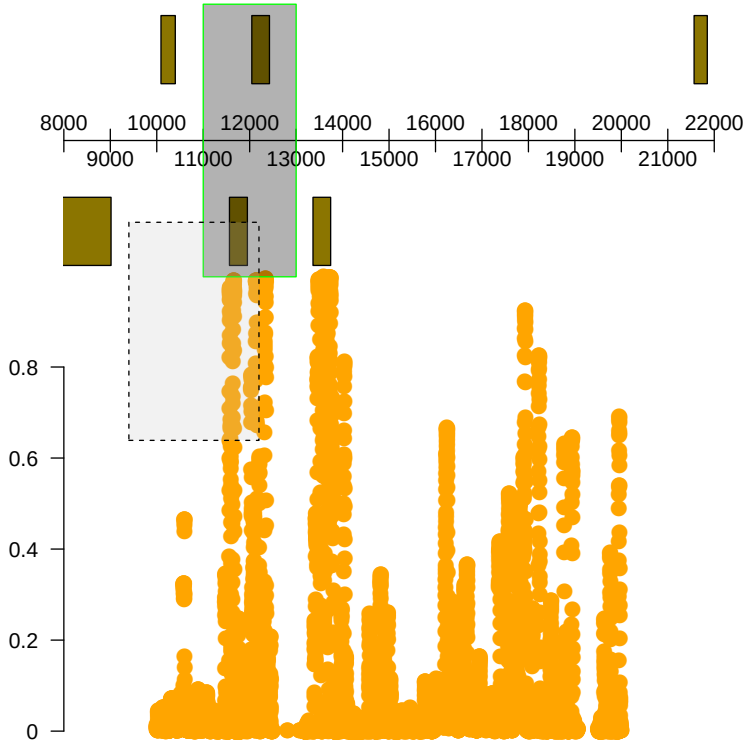
A little nifty feature is to allow alpha blending to make things slightly transparent. If the device you wish to plot on however, does not support transparency then you will get a warning.

```
> ro <- makeRectangleOverlay(start = 11000, end = 13000, region = c(1,3),
+           dp = DisplayPars(color = "green", alpha = .3))
> to <- makeTextOverlay("here is some text", xpos = 15000, ypos = .95)
> gdPlot(list(grF, ga, grR, bt), overlay = c(ro, to))
```



Also, one can use "absolute" coordinates to specify a region just in case one wants to be a bit more precise.

```
> roR <- makeRectangleOverlay(start = .1, end = .3, coords = "absolute",
+                             dp = DisplayPars(fill = "grey", alpha = .2, lty = "dash
+                             region = c(.4,.7))
> gdPlot(list(grF, ga, grR, bt), overlays = list(ro, roR))
```

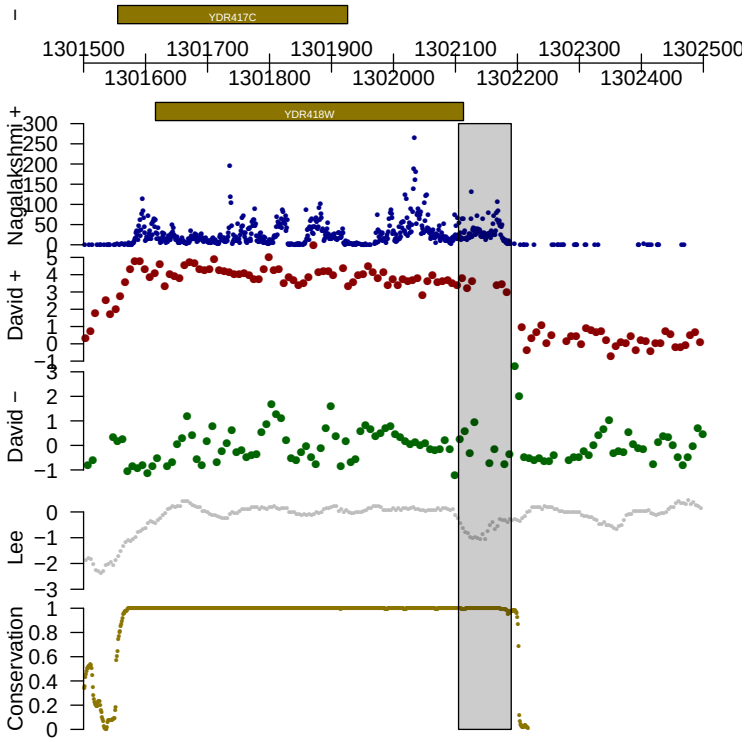


4.2 GenomeGraphs Classes

```
> data("seqDataEx", package = "GenomeGraphs")
> str = seqDataEx$david[,"strand"] == 1
> biomart = useMart("ensembl", "scerevisiae_gene_ensembl")
> pList = list("-" = makeGeneRegion(chromosome = "IV", start = 1300000, end = 1310000
+                                   strand = "-", biomart = biomart,
+                                   dp = DisplayPars(plotId = TRUE, idRotation = 0,
+              makeGenomeAxis(dp = DisplayPars(size = 3)),
+              "+" = makeGeneRegion(chromosome = "IV", start = 1300000, end = 131000
+                                   strand = "+", biomart = biomart,
+                                   dp = DisplayPars(plotId = TRUE, idRotation = 0,
+              "Nagalakshmi" = makeBaseTrack(base = seqDataEx$snyder[, "location"],
+                                            dp = DisplayPars(lwd = .3, color = "dar
+              "David +" = makeGenericArray(probeStart = seqDataEx$david[str, "locat
+                                           intensity = seqDataEx$david[str, "expr",
+                                           dp = DisplayPars(pointSize = .5)),
+              "David -" = makeGenericArray(probeStart = seqDataEx$david[!str, "loca
```

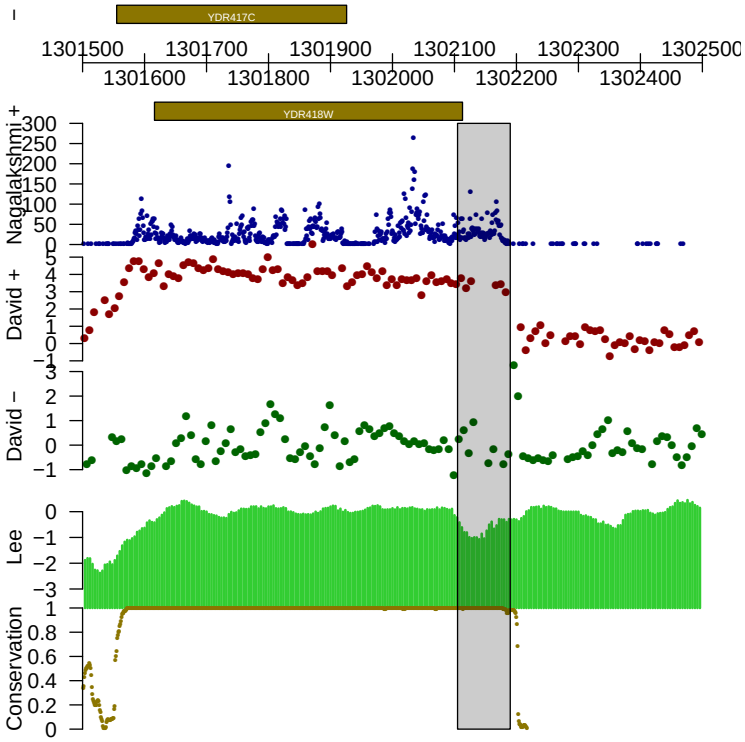
class	description
gdObject	the root class of the system, never directly instantiated
Gene	class representing a gene
GeneRegion	class defining a region of a chromsome, generally a set of genetic elements (genes)
Transcript	class defining a transcript
TranscriptRegion	class defining a region of a chromsome, generally a set of genetic elements (transcripts)
Ideogram	an ideogram
Title	class to draw a title
Legend	class to draw a legend
GenomeAxis	class to draw a axis
Segmentation	class to draw horizontal lines in various sets of data
GenericArray	class to draw data from microarrays.
ExonArray	class to draw data from exon microarrays.
GeneModel	class to draw custom gene models (intron-exon structures)
BaseTrack	class to draw whatever kind of data at a given base
MappedRead	class to plot sequencing reads that are mapped to the genome
DisplayPars	class managing various plotting parameters
AnnotationTrack	class used to represent custom annotation
Overlay	root class for overlays, never directly instantiated
RectangleOverlay	class to represent rectangular regions of interest
TextOverlay	class to draw text on plots

```
+                                                intensity = seqDataEx$david[!str, "expr"
+                  dp = DisplayPars(color = "darkgreen", pointSize = .5)),
+                "Lee" = makeBaseTrack(base = seqDataEx$nislow[, "location"],
+                                      value = seqDataEx$nislow[, "evalue"], dp = Disp
+                "Conservation" = makeBaseTrack(base = seqDataEx$conservation[, "locat
+                                               value = seqDataEx$conservation[, "scor
+                                               dp = DisplayPars(color="gold4", lwd=.2
> gdPlot(pList, minBase = 1301500, maxBase = 1302500,
+        overlay = makeRectangleOverlay(start = 1302105, end = 1302190, region = c(4,
```



We can also employ different plotting types for the BaseTrack object.

```
> setPar(pList$Lee@dp, "type", "h")
> setPar(pList$Lee@dp, "color", "limegreen")
> setPar(pList$Lee@dp, "lwd", 2)
> gdPlot(pList, minBase = 1301500, maxBase = 1302500,
+        overlay = makeRectangleOverlay(start = 1302105, end = 1302190, region = c(4,
+        dp = DisplayPars(alpha = .2)))
```


YDR417C
1301500
1301700
1301900
1302100
1302300
1302500
1301600
1301800
1302000
1302200
1302400
YDR418W
Nagalakshmi +
David +
David −
Lee
Conservation