Today is November 29, 2012.

```
> rm(list = ls())
> library("vcf2R")
> library("GenomicRanges")
> library("BSgenome.Hsapiens.UCSC.hg19")
> library("Gviz")
```

1 Targeted Loci

```
> file <- system.file("extdata", "snvs.fixed_header.frq",
    package = "vcf2R")
> pos.df <- frq2pos(file)
> head(pos.df)

  chr      pos
1   1 18771811
2   1 18771817
3   1 18771849
4   1 18771875
5   1 18771893
6   1 18771920

> with(pos.df, table(chr))

chr
    1    10    14    17    20     4     8     9
44713 29857  1559 29486 18740  2444 27639 20751

> pos.gr <- with(pos.df, GRanges(seqnames = paste("chr",
    chr, sep = ""), ranges = IRanges(start = pos, end = pos),
    strand = "*"))
> seqlengths(pos.gr) <- seqlengths(Hsapiens)[levels(seqnames(pos.gr))]
> start(pos.gr) <- start(pos.gr) - 10000
> end(pos.gr) <- end(pos.gr) + 10000
> loci <- reduce(pos.gr)
> start(loci) <- start(loci) + 10000
> end(loci) <- end(loci) - 10000
```

```
> val <- data.frame(n.snps = countOverlaps(loci, pos.gr),
     kb = width(loci)/1000)
> values(loci) <- val
> names(loci) <- c("PAX7", "ABCA4", "IRF6", "VAX1", "FGFR2",
     "BMP4", "NTN1", "NOG", "MAFB", "MSX1", "8q24", "PTCH1",
     "FOXE1")
> loci

GRanges with 13 ranges and 2 metadata columns:
        seqnames                ranges strand |    n.snps        kb
           <Rle>             <IRanges>  <Rle> | <integer> <numeric>
   PAX7     chr1 [ 18771811,  19208536]      * |     13309   436.726
  ABCA4     chr1 [ 94324232,  95013570]      * |     16310   689.339
   IRF6     chr1 [209836758, 210468863]      * |     15094   632.106
   VAX1    chr10 [118421219, 119167920]      * |     18546   746.702
  FGFR2    chr10 [123095899, 123499252]      * |     11311   403.354
   BMP4    chr14 [ 54382326,  54445461]      * |      1559    63.136
   NTN1    chr17 [  8754615,   9266552]      * |     15212   511.938
    NOG    chr17 [ 54402391,  54957844]      * |     14274   555.454
   MAFB    chr20 [ 38902239,  39614945]      * |     18740   712.707
   MSX1     chr4 [  4824627,   4901842]      * |      2444    77.216
   8q24     chr8 [129295457, 130355340]      * |     27639  1059.884
  PTCH1     chr9 [ 98133217,  98413595]      * |      7795   280.379
  FOXE1     chr9 [100357256, 100877263]      * |     12956   520.008
  ---
  seqlengths:
        chr1     chr10     chr14 ...      chr4      chr8      chr9
   249250621 135534747 107349540 ... 191154276 146364022 141213431

> sum(width(loci))/1e+06

[1] 6.688949

> with(values(loci), sum(n.snps))

[1] 175189

> data("targets.hg19")
> targets.hg19.gr

GRanges with 13 ranges and 0 metadata columns:
        seqnames                ranges strand
           <Rle>             <IRanges>  <Rle>
   IRF6     chr1 [209837199, 210468406]      *
   MAFB    chr20 [ 38902646,  39614513]      *
  ABCA4     chr1 [ 94324660,  95013109]      *
   8q24     chr8 [129295896, 130354946]      *
  FOXE1     chr9 [100357692, 100876841]      *
   PAX7     chr1 [ 18772300,  19208054]      *
   VAX1    chr10 [118421625, 119167424]      *
   NTN1    chr17 [  8755114,   9266060]      *
    NOG    chr17 [ 54402837,  54957390]      *
```

```
 MSX1     chr4 [  4825126,   4901385]      *
 BMP4    chr14 [ 54382690,  54445053]      *
FGFR2    chr10 [123096374, 123498771]      *
PTCH1     chr9 [ 98133647,  98413162]      *
---
seqlengths:
  chr1 chr10 chr14 chr17 chr20  chr4  chr8  chr9
    NA    NA    NA    NA    NA    NA    NA    NA
```

2 Regions

```
> known.gene.file <- system.file("extdata", "known-genes",
    package = "vcf2R")
> kg.df <- data.frame(scan(file = known.gene.file, sep = "\t",
    what = list("", 1L, 1L, "", "", "", 1L, 1L, 1L, 1L,
        "", "")))
> names(kg.df) <- c("chr", "start", "end", "name", "foo",
    "strand", "cds.start", "cds.end", "foo2", "num.exons",
    "exon.coods.start", "exon.coods.end")
> kg.gr <- GRanges(seqnames = kg.df$chr, ranges = IRanges(start = kg.df$start,
    end = kg.df$end), strand = kg.df$strand)

> ht <- 4
> wd <- ht * 1.618
> for (locus in names(loci)) {
    gr <- targets.hg19.gr[names(targets.hg19.gr) == locus,
        ]
    chr <- as.character(unique(seqnames(gr)))
    seqlengths(gr) <- seqlengths(Hsapiens)[levels(seqnames(gr))]
    atrack <- AnnotationTrack(gr, name = paste0(locus,
        " Target"), chromosome = chr, genome = "hg19",
        stacking = "squish")
    atrack2 <- AnnotationTrack(loci[names(loci) == locus,
        ], chromosome = chr, genome = "hg19", name = paste0(locus,
        " Actual"), stacking = "squish")
    gtrack <- GenomeAxisTrack()
    itrack <- IdeogramTrack(genome = "hg19", chromosome = chr)
    kg.sub.df <- as.data.frame(kg.gr[subjectHits(findOverlaps(loci[names(loci) ==
        locus, ], kg.gr)), ])
    if (nrow(kg.sub.df) != 0) {
        kg.sub.df <- kg.sub.df[, -c(1, 4, 5)]
        grtrack <- GeneRegionTrack(kg.sub.df, genome = "hg19",
            chromosome = as.character(seqnames(loci)[which(names(loci) ==
                locus)]), name = "Known Genes")
        pdf(file = paste0("./figures/loci/", locus, ".pdf"),
            height = ht, width = wd)
        plotTracks(list(atrack2, atrack, grtrack, gtrack,
            itrack), add = FALSE, main = locus)
        dev.off()
    }
    else {
```

```
        pdf(file = paste0("./figures/loci/", locus, ".pdf"),
            height = ht, width = wd)
        plotTracks(list(atrack2, atrack, gtrack, itrack),
            add = FALSE, main = locus)
        dev.off()
    }
}
```

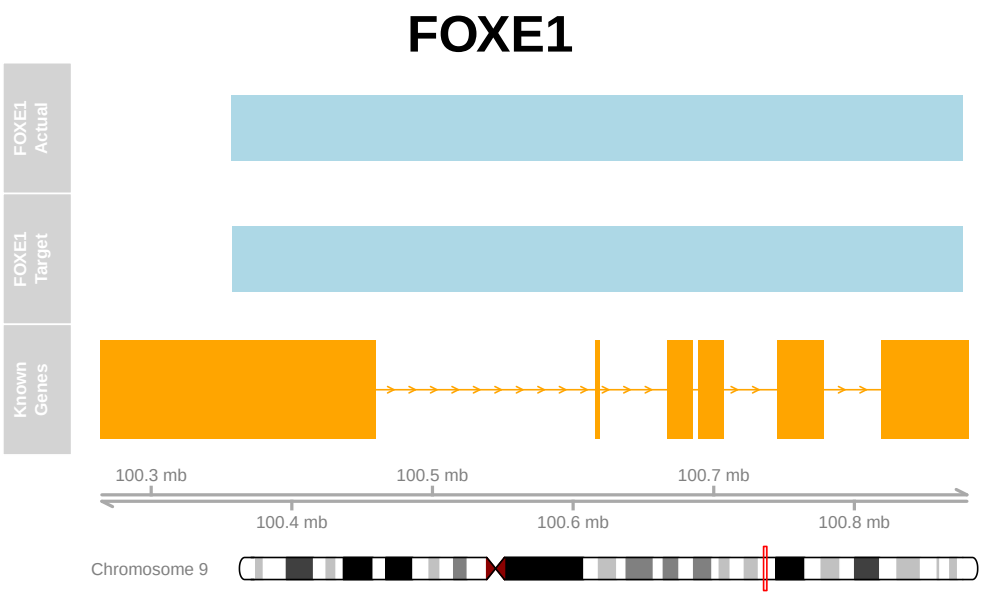
```
> kg.sub.df
```

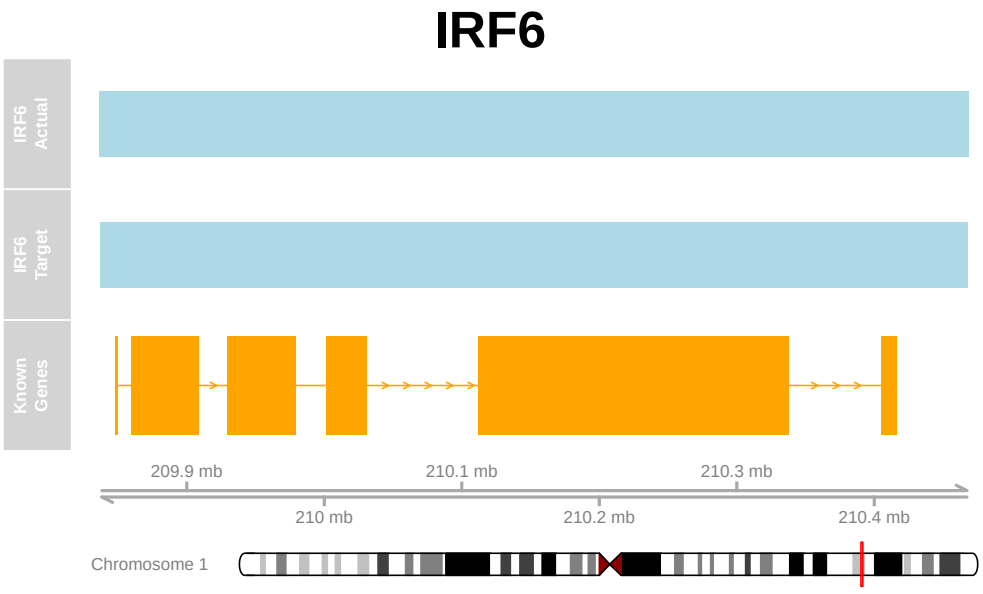
```
       start       end
1  100263461 100364025
2  100263920 100364025
3  100365296 100389046
4  100362361 100395962
5  100362361 100395962
6  100395704 100436029
7  100440044 100441883
8  100437190 100459691
9  100437190 100459691
10 100615536 100618997
11 100666771 100684852
12 100666771 100684852
13 100672241 100684852
14 100673199 100684852
15 100689072 100700526
16 100689072 100707134
17 100745488 100778224
18 100840474 100844448
19 100818958 100845365
20 100846636 100854843
21 100846636 100854843
22 100831568 100881488
23 100846636 100881488
24 100846636 100881488
```

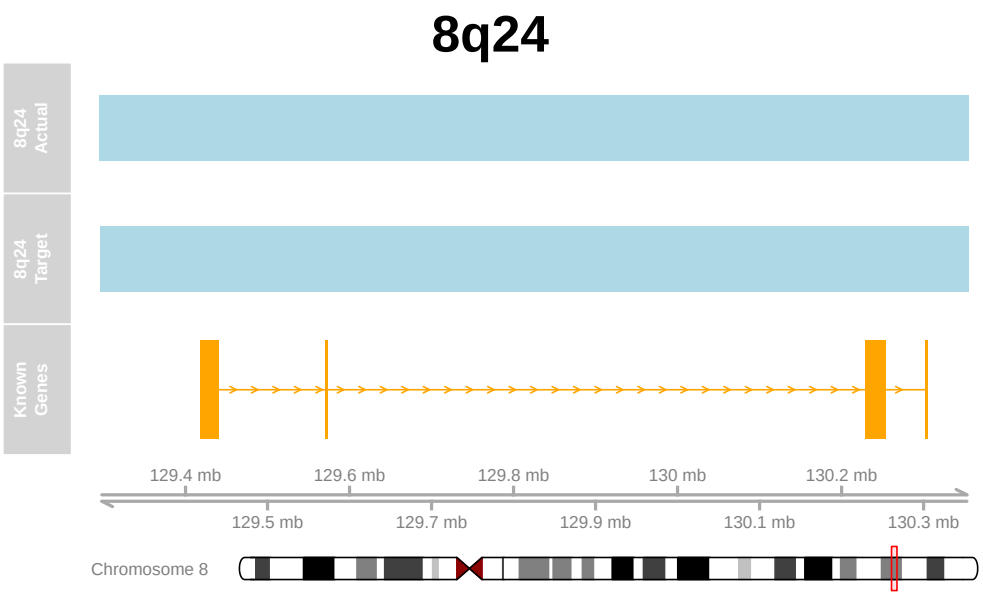
3 Hardy-Weinberg Equilibrium

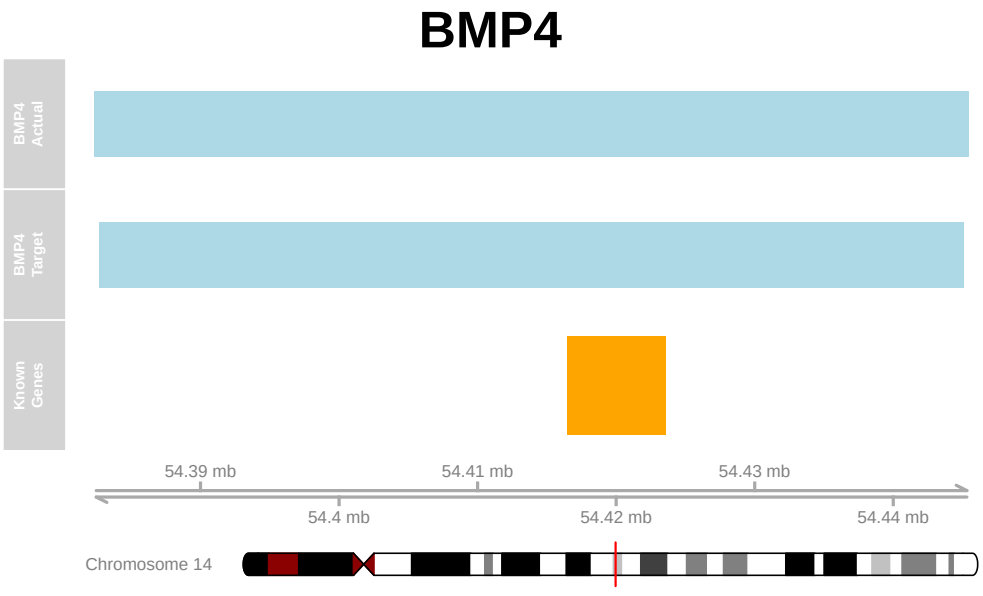
```
> file <- system.file("extdata", "snvs.fixed_header.hwe",
    package = "vcf2R")
> hwe.df <- hwe2R(file = file)
> head(hwe.df)
```

```
  chr      pos    chi.sq  p.value homo.major het homo.minor
1   1 18772169  0.000056 1.000000       4494   1          0
2   1 18772190  0.000223 1.000000       4493   2          0
3   1 18772191 15.727310 0.053862       4464  30          1
4   1 18772203  0.000223 1.000000       4493   2          0
5   1 18772218  0.000223 1.000000       4493   2          0
6   1 18772236  0.016134 1.000000       4478  17          0
```


FOXE1
FOXE1 Actual
FOXE1 Target
Known Genes
100.3 mb
100.5 mb
100.7 mb
100.4 mb
100.6 mb
100.8 mb
Chromosome 9


IRF6
IRF6 Actual
IRF6 Target
Known Genes
209.9 mb
210.1 mb
210.3 mb
210 mb
210.2 mb
210.4 mb
Chromosome 1


8q24
8q24 Actual
8q24 Target
Known Genes
129.4 mb
129.6 mb
129.8 mb
130 mb
130.2 mb
129.5 mb
129.7 mb
129.9 mb
130.1 mb
130.3 mb
Chromosome 8


BMP4
BMP4 Actual
BMP4 Target
Known Genes
54.39 mb
54.41 mb
54.43 mb
54.4 mb
54.42 mb
54.44 mb
Chromosome 14