

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

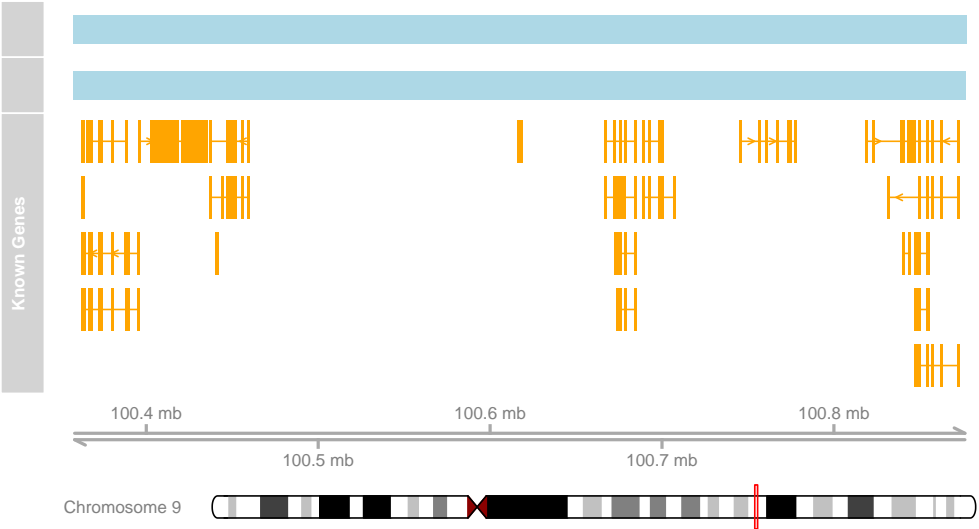
```

> data(kg.hg19)
> kg.gr <- GRanges(seqnames = kg.hg19$chr, ranges = IRanges(start = kg.hg19$start,
  end = kg.hg19$end), strand = kg.hg19$strand, exon = as.factor(kg.hg19$exon),
  gene = as.factor(gene <- substr(kg.hg19$exon, 1,
    8)), feature = as.factor(gene), transcript = as.factor(gene),
  symbol = as.factor(gene), id = as.factor(gene))

> 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.gr <- kg.gr[subjectHits(findOverlaps(loci[names(loci) ==
    locus, ], kg.gr)), ]
  if (length(kg.sub.gr) != 0) {
    grtrack <- GeneRegionTrack(range = kg.sub.gr,
      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()
  }
}

```

# FOXE1



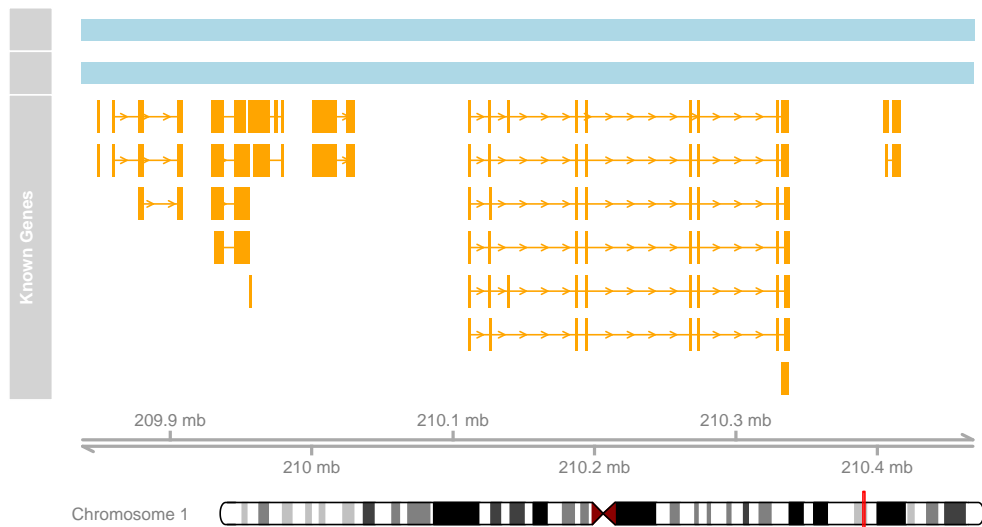
```
}
}
```

## 3 Hardy-Weinberg Equilibrium

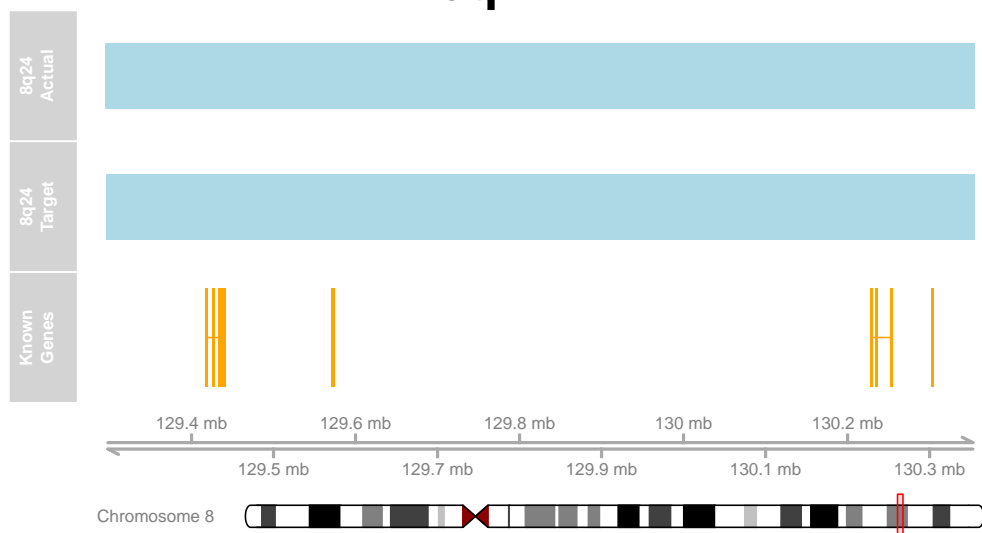
```
> file <- system.file("extdata", "snvs.fixed_header.hwe",
  package = "vcf2R")
> hwe.df <- hwe2R(file = file)
> head(hwe.df)
```

|   | 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          |

# IRF6



# 8q24



# BMP4

