Today is November 26, 2012.

```
> library("vcf2R")
> library("GenomicRanges")
> library("BSgenome.Hsapiens.UCSC.hg19")
```

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
```

2 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
```