

Week 6 Lab Session

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Section 1. Proportion of G/G in a population

Downloaded a CSV file from Ensembl https://useast.ensembl.org/Homo_sapiens/Variation/Sample?db=core;r=17:39894595-39895595;v=rs8067378;vdb=variation;vf=959672880#373531_tablePanel

Here we read this CSV file

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
head(mxl)
```

```
## Sample..Male.Female.Unknown. Genotype..forward.strand. Population.s. Father
## 1 NA19648 (F) A|A ALL, AMR, MXL -
## 2 NA19649 (M) G|G ALL, AMR, MXL -
## 3 NA19651 (F) A|A ALL, AMR, MXL -
## 4 NA19652 (M) G|G ALL, AMR, MXL -
## 5 NA19654 (F) G|G ALL, AMR, MXL -
## 6 NA19655 (M) A|G ALL, AMR, MXL -
## Mother
## 1 -
## 2 -
## 3 -
## 4 -
## 5 -
## 6 -
```

```
table(mxl$Genotype..forward.strand.)
```

```
##
## A|A A|G G|A G|G
## 22 21 12 9
```

```
table(mxl$Genotype..forward.strand.)/nrow(mxl)*100
```

```
##
## A|A A|G G|A G|G
## 34.3750 32.8125 18.7500 14.0625
```

Now let's look at a different population. I picked the GBR.

```
gbr <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
```

Find proportion of G|G

```
round(table(gbr$Genotype..forward.strand.)/nrow(gbr)*100, 2)
```

```
##
## A|A A|G G|A G|G
```

```
## 25.27 18.68 26.37 29.67
```

This variant that is associated with childhood asthma is more frequent in the GBR population than the MXL population.

Let's now dig into this further.

Section 4: Population Scale Analysis [OPTIONAL]

One sample is obviously not enough to know what is happening in a population. You are interested in assessing genetic differences on a population scale. So, you processed about ~230 samples and did the normalization on a genome level. Now, you want to find whether there is any association of the 4 asthma-associated SNPs (rs8067378...) on ORMDL3 expression.

How many samples do we have?

```
expr <- read.table("genotype.txt")
head(expr)
```

```
##      sample geno      exp
## 1 HG00367   A/G 28.96038
## 2 NA20768   A/G 20.24449
## 3 HG00361   A/A 31.32628
## 4 HG00135   A/A 34.11169
## 5 NA18870   G/G 18.25141
## 6 NA11993   A/A 32.89721
```

```
nrow(expr)
```

```
## [1] 462
```

```
table(expr$geno)
```

```
##
## A/A A/G G/G
## 108 233 121
```

```
library(ggplot2)
```

Let's make a box plot

```
ggplot(expr) +
  aes(geno, exp, fill=geno) +
  geom_boxplot(notch=TRUE)
```

