Class 12: Introduction to Genome Informatics Lab

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2024-02-21

Section 1. Proportion og G/G in population

```
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
```

Let's look at different population (GBR)

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

Find proportion of G|G

```r
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

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.

How many samples do we have? > Q13: Read this file into R and determine the sample size for each genotype and their corresponding median expression levels for each of these genotypes.

```r
expr <- read.table("rs8067378_ENSG00000172057.6.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
```

```r
nrow(expr)
```

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

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

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

> The sample sixe for A/A is 108, 233 for A/G, and 121 for G/G.

```r
summary(expr)
```

```
##     sample              geno                exp
##  Length:462         Length:462         Min.   : 6.675
##  Class :character   Class :character   1st Qu.:20.004
##  Mode  :character   Mode  :character   Median :25.116
##                                        Mean   :25.640
##                                        3rd Qu.:30.779
##                                        Max.   :51.518
```

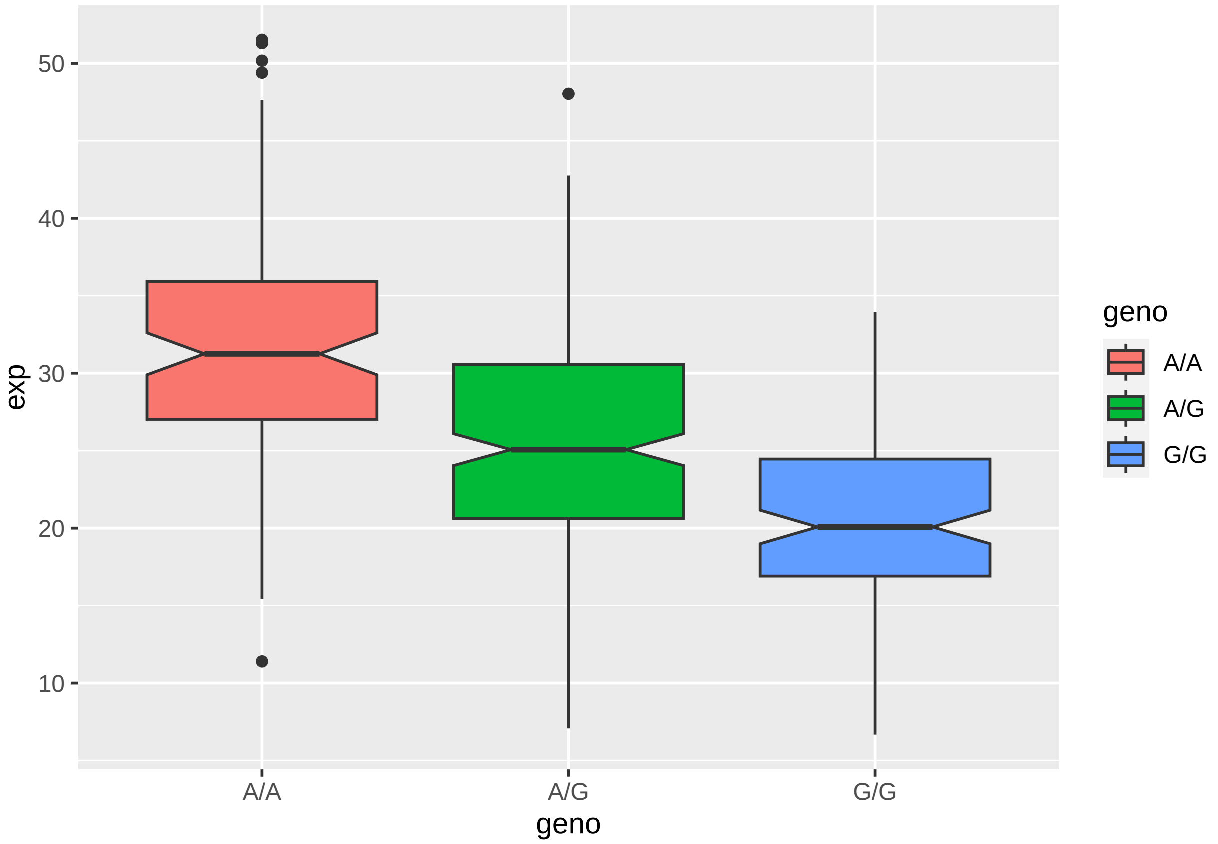
The median expression level is 25.116.

Q14: Generate a boxplot with a box per genotype, what could you infer from the relative expression value between A/A and G/G displayed in this plot? Does the SNP effect the expression of ORMDL3?

```
library(ggplot2)
```

Let's make a boxplot

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



> The expression for G/G tends to be underexpressed/lower than expression for A/A. It is possible that the SNP affects the expression of ORMDL3.