Homework 11

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You are interested in assessing genetic differences on a population scale. So, you processed about $\sim\!230$ 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.

```
expression = read.table("https://marcos-diazg.github.io/BIMM143_SP23/class-material/class1
summary(expression)
```

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

```
median(expression$exp[expression$geno == "A/A"])
```

```
[1] 31.24847
```

```
median(expression$exp[expression$geno == "A/G"])
```

[1] 25.06486

```
median(expression$exp[expression$geno == "G/G"])
```

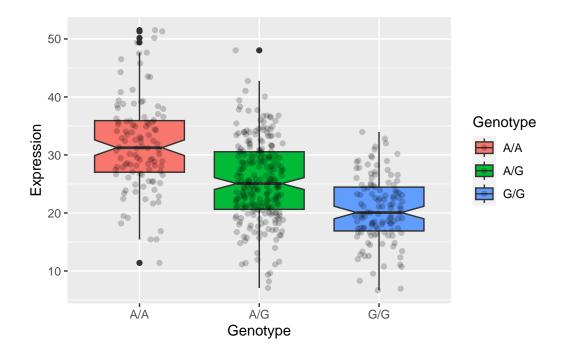
[1] 20.07363

```
table(expression$geno)
```

A/A A/G G/G 108 233 121

Q13: The sample size for each genotype is: 108 samples for the A/A genotype, 233 for A/G, and 121 for G/G. The median of each genotype is 31.2 (A/A), 25.1 (A/G), and 20.1 (G/G).

```
library(ggplot2)
ggplot(expression) + aes(geno, exp, fill=geno) +
  geom_boxplot(notch = TRUE) +
  geom_jitter(color="black", alpha=0.2, width = 0.2) +
  xlab("Genotype") + ylab("Expression") + labs(fill = "Genotype")
```



Q14: From the graph, A/A has higher expression on ORMDL3 than G/G genotype. The genotype difference is correlated to the expression differences, which indicates the SNP might affect the ORMDL3 expression.