## **Lab** 11

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## **Section 4 Population Scale Analysis**

First download the file fromm https://bioboot.github.io/bggn213\_W19/class-material/rs8067378\_ENSG000001 into the project directory. Then read the file.

```
url <- "rs8067378_ENSG00000172057.6.txt"
  data <- read.table(url)</pre>
  head(data)
   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
     Q13 What is the sample size for each genotype and their median expression levels?
  nrow(data) #total size of sample
[1] 462
  table(data$geno) #sample size of each genotype
A/A A/G G/G
108 233 121
```

```
#separating the genotypes into individual vectors to find median expression
aa <- data[data$geno == "A/A",]
ag <- data[data$geno == "A/G",]
gg <- data[data$geno == "G/G",]

#two different ways of getting median expression if already separated
median(aa$exp)

[1] 31.24847

median(data[data$geno == "A/G", "exp"])

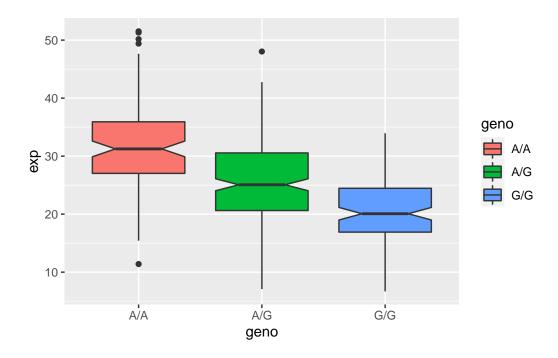
[1] 25.06486

median(gg$exp)

[1] 20.07363

library(ggplot2)

ggplot(data, aes(geno,exp, fill = geno)) +
geom_boxplot(notch = T)</pre>
```



Q14 What could you infer from the real tive expression value between A/A and G/G displayed in this plot? does the SNP effect expression of ORMDL3

A/A has a higher expression than G/G so the G/G genotype has reduced expression of this gene. Depending on the SNP, the expression of ORMDL3 changes; it is higher for A's and lower for G's.