Class 19: Genome informatics

Andrew Kapinos

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> 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.

```
data <- read.table("rs8067378_ENSG00000172057.6.txt")
table(data$geno)
```

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

```
print(paste("The sample size for genotype A/A is", table(data$geno)["A/A"]))
```

```
## [1] "The sample size for genotype A/A is 108"
```

```
print(paste("The sample size for genotype A/G is", table(data$geno)["A/G"]))
```

```
## [1] "The sample size for genotype A/G is 233"
```

```
print(paste("The sample size for genotype G/G is", table(data$geno)["G/G"]))
```

```
## [1] "The sample size for genotype G/G is 121"
```

```
library(dplyr)
summary(data[data$geno=="A/A",]$exp)
```

```
##    Min. 1st Qu.  Median    Mean 3rd Qu.    Max.
##   11.40   27.02   31.25   31.82   35.92   51.52
```

```
print(paste("The median expression value for genotype A/A is",
            round(summary(data[data$geno=="A/A",]$exp)["Median"],3)))
```

```
## [1] "The median expression value for genotype A/A is 31.248"
```

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

```
##    Min. 1st Qu.  Median    Mean 3rd Qu.    Max.
##   7.075  20.626  25.065  25.397  30.552  48.034
```

```
print(paste("The median expression value for genotype A/G is",
            round(summary(data[data$geno=="A/G",]$exp)["Median"],3)))
```

```
## [1] "The median expression value for genotype A/G is 25.065"
```

```
summary(data[data$geno=="G/G",]$exp)
```

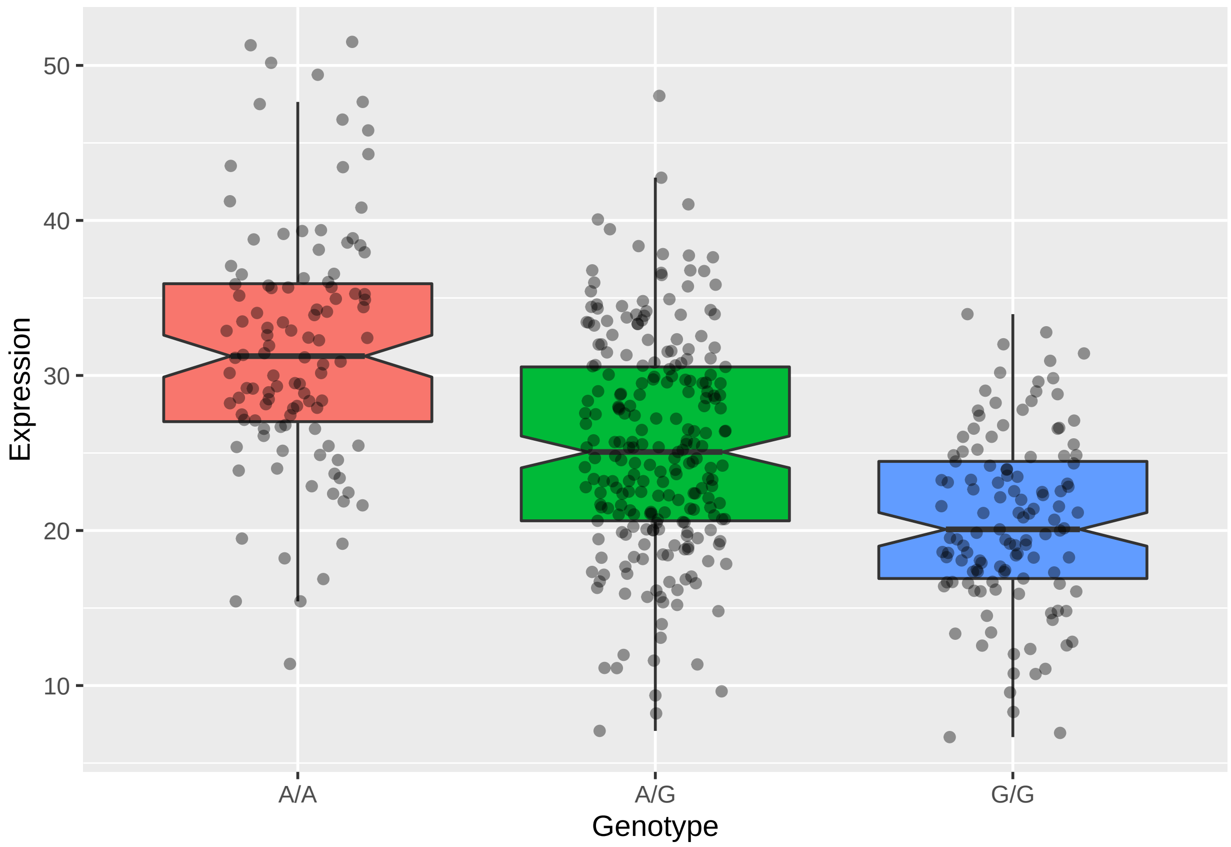
```
##    Min. 1st Qu.  Median    Mean 3rd Qu.    Max.
##   6.675  16.903  20.074  20.594  24.457  33.956
```

```
print(paste("The median expression value for genotype G/G is",
            round(summary(data[data$geno=="G/G",]$exp)["Median"],3)))
```

```
## [1] "The median expression value for genotype G/G is 20.074"
```

> 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)
ggplot(data,aes(geno,exp)) +
  geom_boxplot(notch=TRUE,aes(fill=geno),outlier.shape=NA) +
  geom_point(alpha=0.4,position=position_jitter(0.2)) +
  labs(x="Genotype", y="Expression") +
  theme(legend.position="none")
```



```
t.test(data[data$geno=="A/A",]$exp,data[data$geno=="G/G",]$exp)
```

```
##
##  Welch Two Sample t-test
##
## data:  data[data$geno == "A/A", ]$exp and data[data$geno == "G/G", ]$exp
## t = 12.214, df = 191.65, p-value < 2.2e-16
## alternative hypothesis: true difference in means is not equal to 0
## 95 percent confidence interval:
##   9.412243 13.037619
## sample estimates:
## mean of x mean of y
##  31.81864  20.59371
```

On average, the A/A genotype results in **relatively higher** expression levels when compared to the G/G genotype. Given the results of the boxplot, it would appear as though the SNP does in fact affect expression of ORMDL3.

A Student's t-test confirms that the difference in means is statistically significant, with a p-value of 2.2e-16.