Class 11 Part 2 Population analysis

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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. https://bioboot.github.io/bggn213_W19/class- material/rs8067378_ENSG00000172057.6.txt This is the final file you got (column is genotype and the third column are the expression values.). The first column is sample name, the second

> 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

Importing the data

```
data <- read.table("rs8067378_ENSG00000172057.6.txt", sep = " ")
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
```

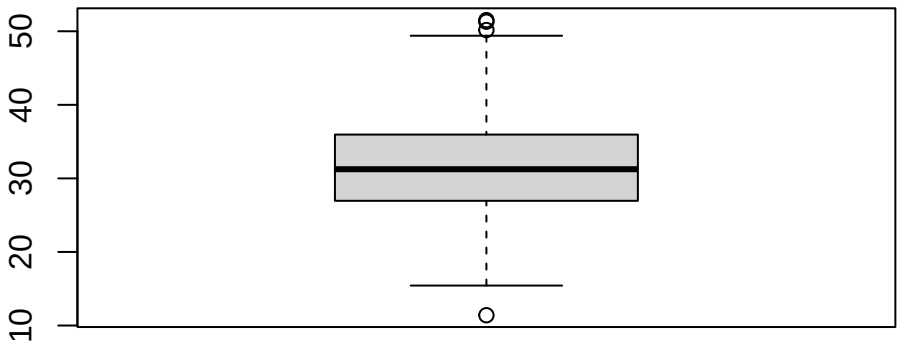
The sample size for each genotype

```
table(data$geno)
```

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

Corresponding median expression levels for each of these genotypes

```
boxinfo <- boxplot(data[data$geno == "A/A", ]$exp)
```



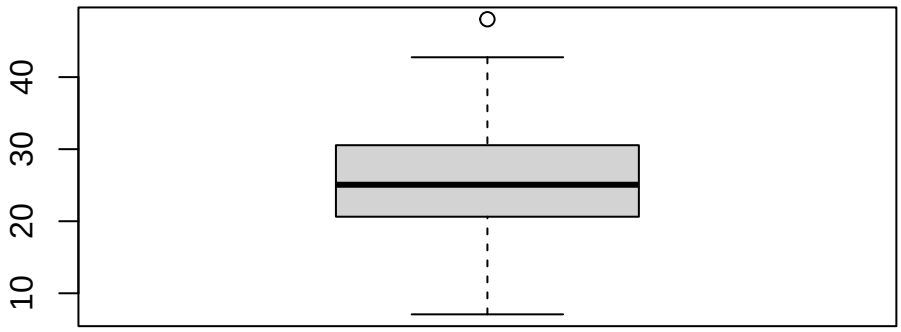

```
medium_val <- boxinfo$stats[3]

medium_val
```

```
[1] 31.24847
```

A/A: 31.24847

```
boxinfo <- boxplot(data[data$geno == "A/G", ]$exp)
```



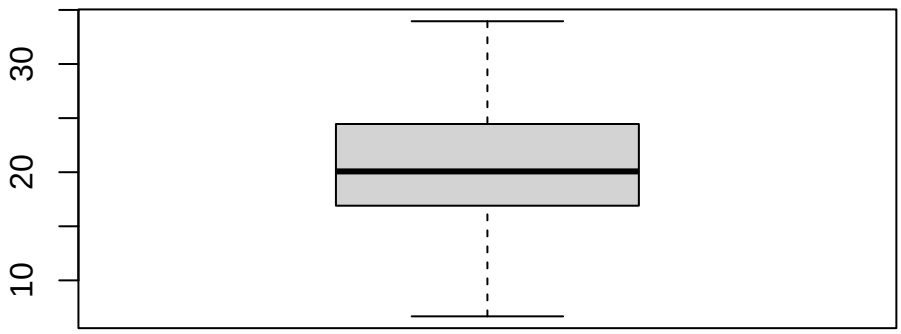

```
medium_val <- boxinfo$stats[3]

medium_val
```

```
[1] 25.06486
```

A/G: 25.06486

```
boxinfo <- boxplot(data[data$geno == "G/G", ]$exp)
```




```
medium_val <- boxinfo$stats[3]

medium_val
```

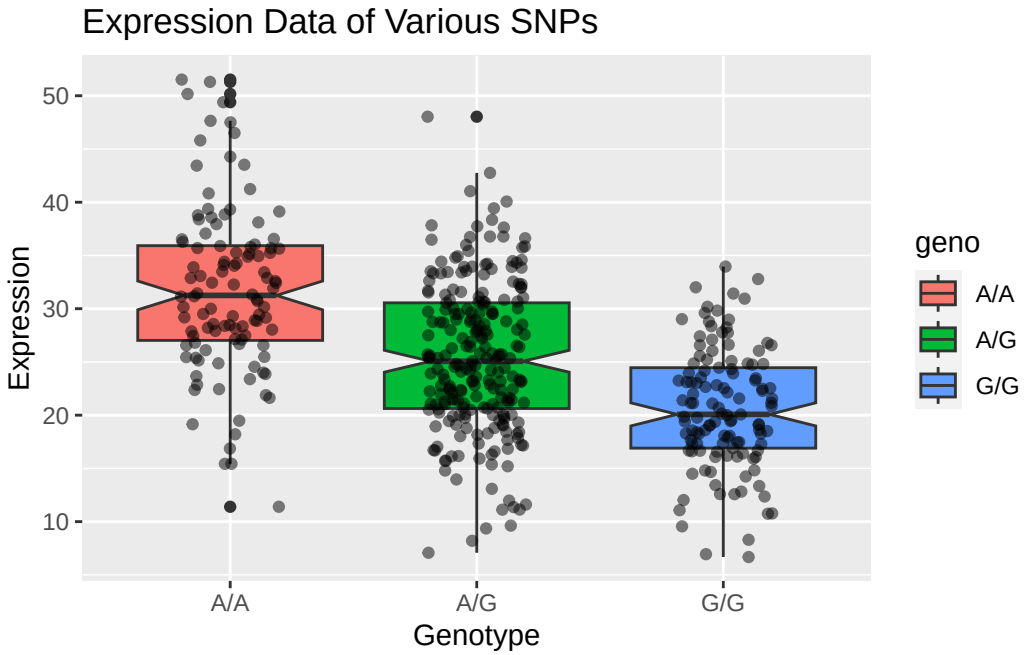
```
[1] 20.07363
```

G/G: 20.07363

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)) +
  geom_jitter(width = 0.2, alpha=0.5) +
  labs(title = "Expression Data of Various SNPs", x = "Genotype", y = "Expression")
```



From the boxplot, I can infer that A/A has relatively greater expression value than G/G, as the median expression of A/A is greater than that of G/G. Since the notches between the boxplots of A/A and G/G do not overlap, this difference is statistically different, suggesting that the SNP does effect the expression of ORMDL3.