class19

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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.

Now, I want to find whether there is any association of the 4 asthma-associated SNPs (rs8067378...) on ORMDL3 expression.

> 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.

```
expr <- read.table("rs8067378_ENSG00000172057.6.txt")
```

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

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

The sample sizes for each genotypes are 108(A/A), 233(A/G), and 121(G/G).

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

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

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

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

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

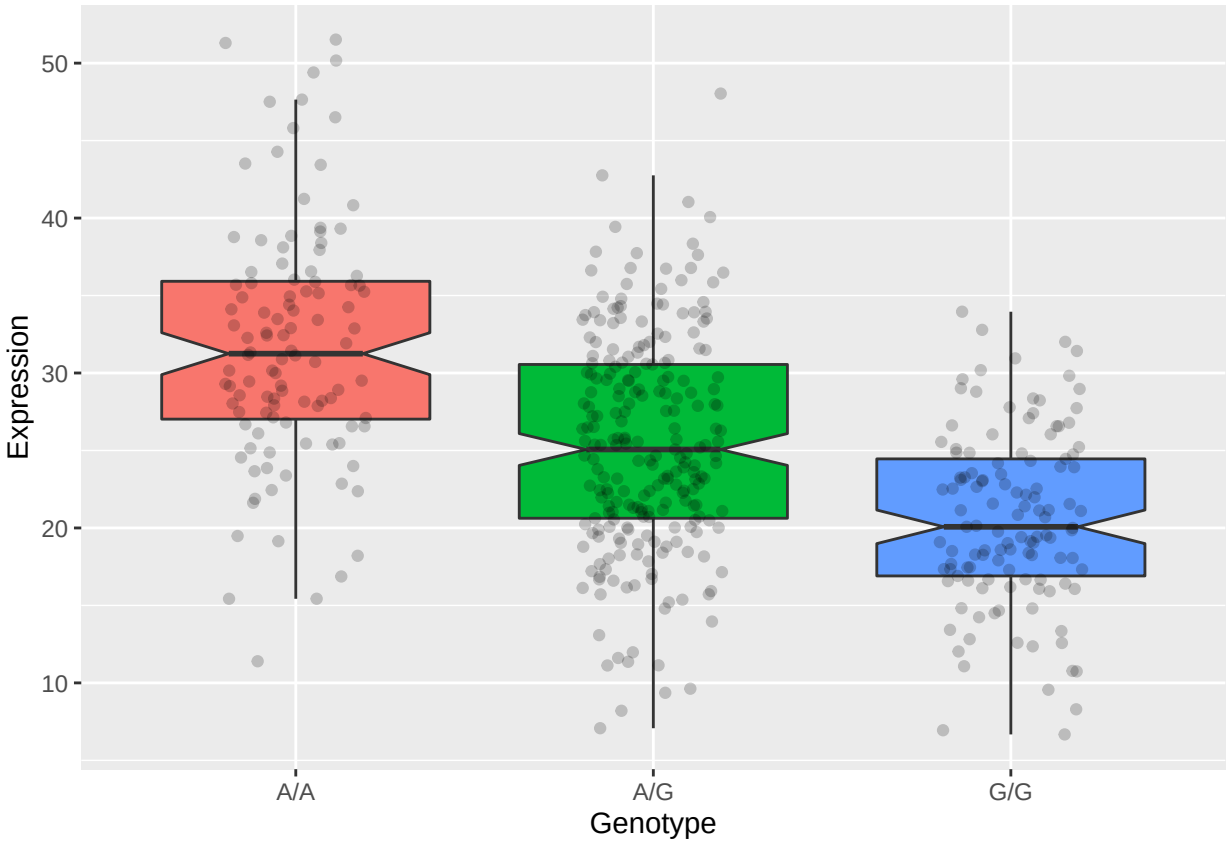
```
##    Min. 1st Qu.  Median    Mean 3rd Qu.    Max.
##   6.675  16.903  20.074  20.594  24.457  33.956
```

The median expression levels for each of these genotypes are 31.25(A/A), 25.065(A/G), and 20.074(G/G).

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(expr) + aes(geno, exp, fill=geno) +
  geom_boxplot(outlier.shape=NA, notch=TRUE) +
  geom_jitter(width=0.2, alpha=0.2) +
  xlab("Genotype") + ylab("Expression") +
  theme(legend.position="none")
```



I can see clear difference in expression value between A/A and G/G. Based on the observation, I think that having a G/G is associated with the reduced expression of the gene.