Population Scale Analysis HW

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We will be taking a look at genetic differences at a population level and see if there's an association between the asthma related SNPs on ORMDL3.

> 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.

Let's see how many samples we have:

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

```
   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
```

```
nrow(expr)
```

```
[1] 462
```

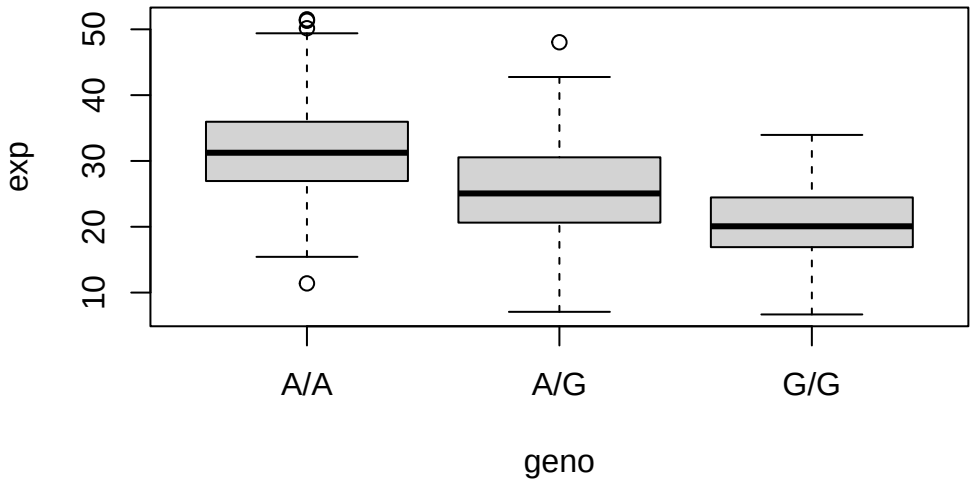
```
table(expr$geno)
```

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

```
summary(expr)
```

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

```
# save the boxplot information into variable medExp
medExp <- boxplot(exp~geno, data = expr)
```




```
medExp$stats
```

```
         [,1]     [,2]     [,3]
[1,] 15.42908  7.07505  6.67482
[2,] 26.95022 20.62572 16.90256
[3,] 31.24847 25.06486 20.07363
```

```
[4,] 35.95503 30.55183 24.45672
[5,] 49.39612 42.75662 33.95602
```

```
medExp$stats[3,]
```

```
[1] 31.24847 25.06486 20.07363
```

The sample size for A/A genotype is 108. A/G has 233 samples and G/G has 121 samples.

The third row of the stats of the boxplot reflect the median of each genotype. The medians go as followed: 31.25 for A/A, 25.06 for A/G, 20.07 for G/G.

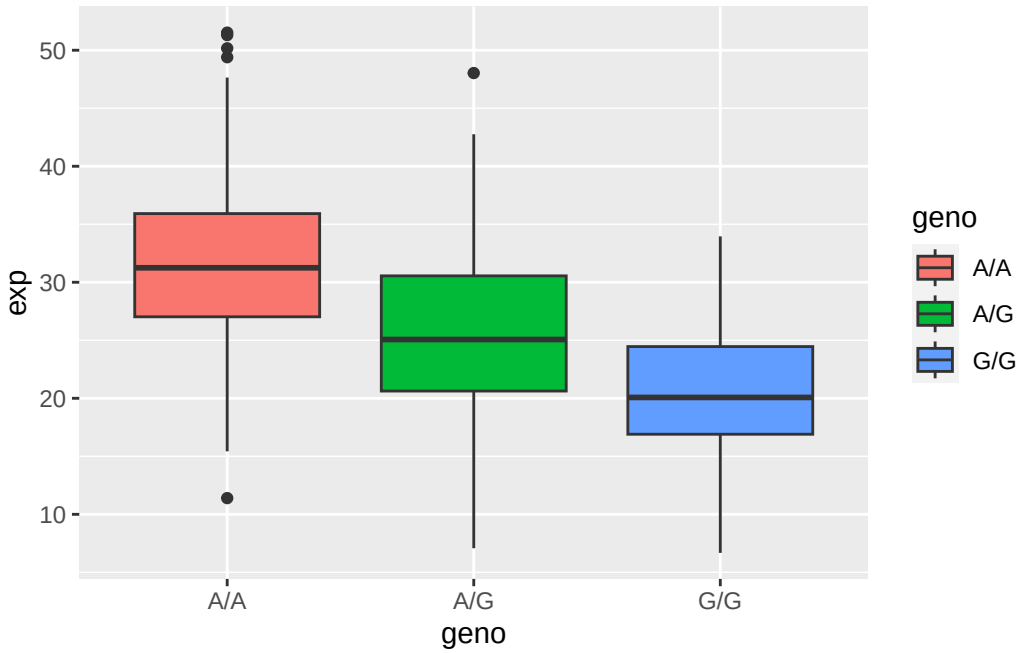
> Q14: Generate a boxplot with a box per genotype, what could you infer from the relative expression value between A/A and G/G displayed in this plot? Does the SNP effect the expression of ORMDL3?

```
library(ggplot2)
```

Let's make a boxplot of our data:

```
# notch adds a belt
ggplot(expr) + aes(x=geno, y=exp, fill = geno) +
  geom_boxplot(noth=TRUE)
```

```
Warning in geom_boxplot(noth = TRUE): Ignoring unknown parameters: `noth`
```



From the plot, we can see that A/A has a higher expression value than G/G. Yes, the SNP affects the expression of ORMDL3 as the genotype for G/G is reduced with its lower median.