## Class 12: Population Scale Analysis [HOMEWORK]

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##Section 4: Population Scale Analysis [HOMEWORK]
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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  $\sim 230$  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.

How many samples do we have? **Answer:** 462

```
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

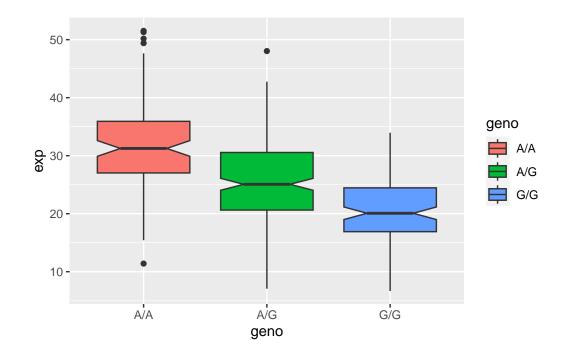
Q13. Determine the sample size for each genotype:
table(expr$geno)
```

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

```
library(ggplot2)
```

Lets make a boxplot.

```
ggplot(expr) + aes(x=geno, y=exp, fill=geno) +
geom_boxplot(notch=TRUE)
```



Q14. 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? **Answer:** Having a G/G is associated with having a reduced expression of the gene. And the SNP does affect the expression of ORMDL3.