

Class11

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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. 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.

How many samples do we have?

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")
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
```

We have 462 samples

```
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

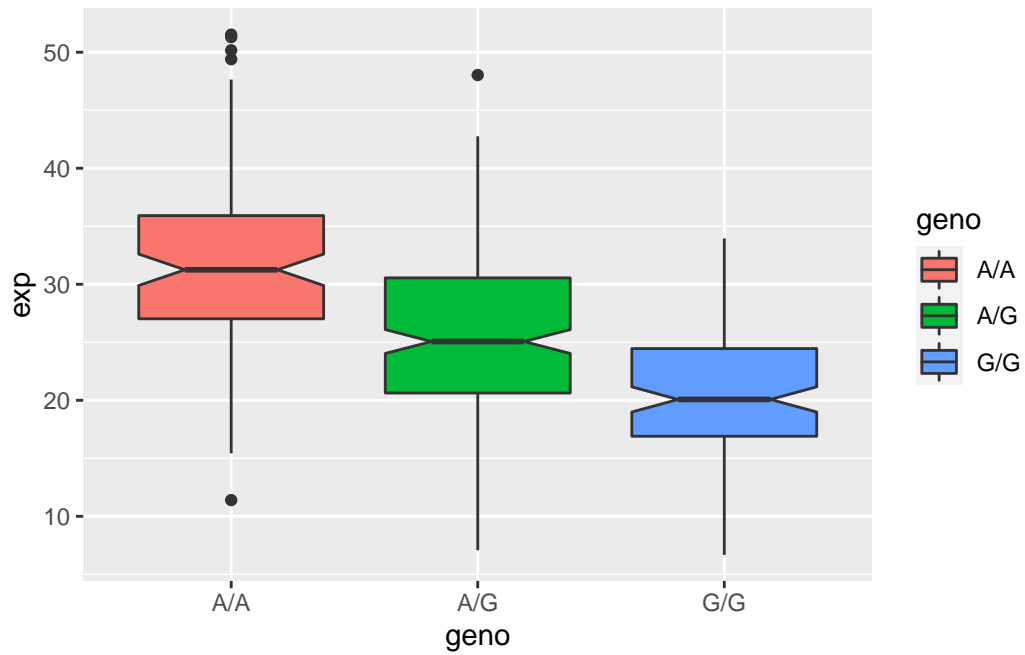
There is a median of 25.116

```
library(ggplot2)
```

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?

Let's make a boxplot

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



The SNP does effect the expression of ORMDL3. We can see that having a G/G in this location is associated with having a reduced expression of this gene.