

Section 4: Homework

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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?

```
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
```

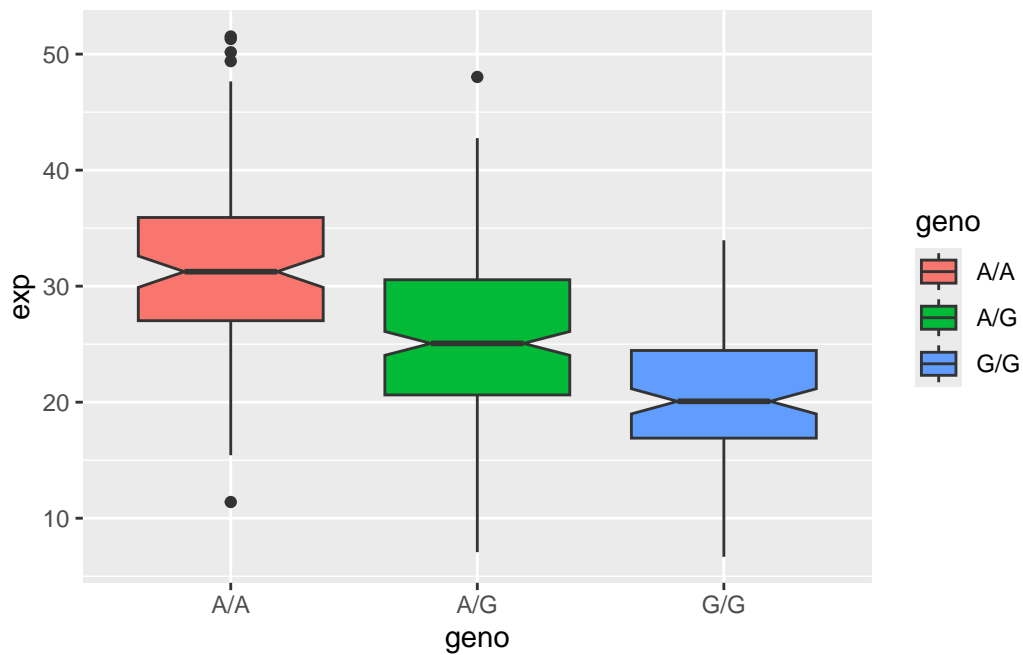
```
table(expr$geno)
```

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

```
library(ggplot2)
```

Lets make a box plot

```
boxplot <- ggplot(expr)+  
  aes(geno,exp,fill=geno)+  
  geom_boxplot(notch=T)  
boxplot
```



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.

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

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

```
library(dplyr)
```

Attaching package: 'dplyr'

The following objects are masked from 'package:stats':

filter, lag

The following objects are masked from 'package:base':

intersect, setdiff, setequal, union

```
medians <- expr %>%  
  group_by(geno) %>%  
  summarize(median_exp = median(exp))  
medians
```

```
# A tibble: 3 x 2  
  geno median_exp  
  <chr>      <dbl>  
1 A/A         31.2  
2 A/G         25.1  
3 G/G         20.1
```

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 ORM DL3?

People with G/G genotype have a lower expression level of ORM DL3 compared to people with A/A genotype. SNP do affect the expression of ORM DL3.