week8 HW

Ruofan Kang (A172369210)

Section 1. Proportion og G/G in a population

Downloaded a CSV file from Ensemble < https://www.ensembl.org/Homo_sapiens/Variation/Sample?db=core;
39895595;v=rs8067378;vdb=variation;vf=105535077#373531_tablePanel

Here we read this CSV file

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
head(mxl)
```

```
  Sample..Male.Female.Unknown. Genotype..forward.strand. Population.s. Father
1                 NA19648 (F)                         A|A ALL, AMR, MXL      -
2                 NA19649 (M)                         G|G ALL, AMR, MXL      -
3                 NA19651 (F)                         A|A ALL, AMR, MXL      -
4                 NA19652 (M)                         G|G ALL, AMR, MXL      -
5                 NA19654 (F)                         G|G ALL, AMR, MXL      -
6                 NA19655 (M)                         A|G ALL, AMR, MXL      -
  Mother
1      -
2      -
3      -
4      -
5      -
6      -
```

```
table(mxl$Genotype..forward.strand.)
```

```
A|A A|G G|A G|G
 22  21  12   9
```

```
table(mxl$Genotype..forward.strand.)/nrow(mxl)*100
```

```
    A|A     A|G     G|A     G|G
34.3750 32.8125 18.7500 14.0625
```

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.

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
```

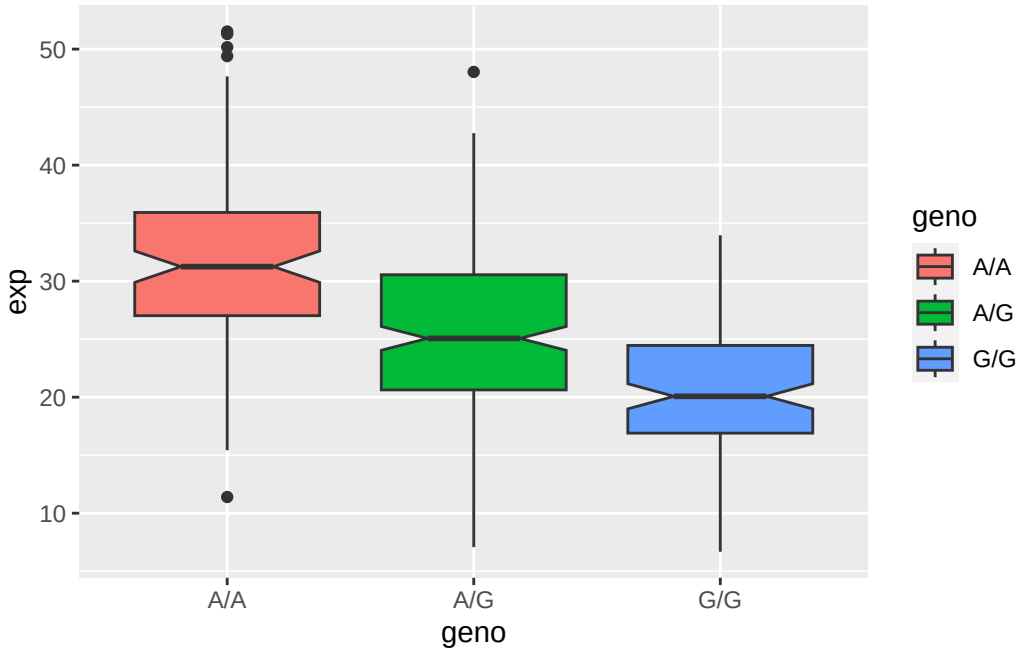
```
table(expr$geno)
```

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

```
library(ggplot2)
```

Lets make a boxplot

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



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?

From the graph, it is observed that individuals with the A/A genotype exhibit higher expression levels of the associated genes, followed by those with the A/G genotype, and then G/G. The relative expression values suggest that the A allele may be linked to higher expression compared to the G allele. Additionally, there are outliers within the A/A and A/G genotype groups, indicating that some samples have expression levels significantly higher or lower than those of the rest of the group. The boxplots imply may have a effect of the SNP on the expression of ORMDL3.