Class12

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Section 1 - Proportion of G/G in Population

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

Q5. What proportion of the Mexican Ancestry in Los Angeles sample population (MXL) are homozygous for the asthma associated SNP (G|G)?

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

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

9 out of 64 (14.01%) of the samples of Mexican ancestry individuals are homozygous G/G

Let's now compare it with the GBR population

```
GBR <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
```

Proportion of G|G in this GBR population is:

```
round(table(GBR$Genotype..forward.strand.)/nrow(GBR)*100, 2)
```

```
  A|A   A|G   G|A   G|G
25.27 18.68 26.37 29.67
```

Here, the proportion of homozygous G in the population is: 29.67%. It is a higher proportion — almost twice more homozygous GG than the MExican nacestry in LA.

HOMEWORK CLASS 12

Section 4: Population Scale Analysis

The following file will be used to determine if the SNP rs8067378 has any effect on the expression of ORMDL3 gene. It shows the genotype of the SNP and the expression level of 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.

Opening the file:

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

Genotypes sample size:

```
table(snpfile$geno)
```

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

Median expression levels for each of the genotypes:

```
#for Genotype A/A, the median expression level of ORMDL3 is 31.25
summary(snpfile[snpfile$geno=="A/A", ])
```

```
   sample              geno                exp
 Length:108         Length:108         Min.   :11.40
 Class :character   Class :character   1st Qu.:27.02
 Mode  :character   Mode  :character   Median :31.25
                                       Mean   :31.82
                                       3rd Qu.:35.92
                                       Max.   :51.52
```

```
#for Genotype A/G, the median expression level of ORMDL3 is 25.07
summary(snpfile[snpfile$geno=="A/G", ])
```

```
   sample              geno                exp
 Length:233         Length:233         Min.   : 7.075
 Class :character   Class :character   1st Qu.:20.626
 Mode  :character   Mode  :character   Median :25.065
                                       Mean   :25.397
                                       3rd Qu.:30.552
                                       Max.   :48.034
```

```
#for Genotype G/G, the median expression level of ORMDL3 is 20.07
summary(snpfile[snpfile$geno=="G/G", ])
```

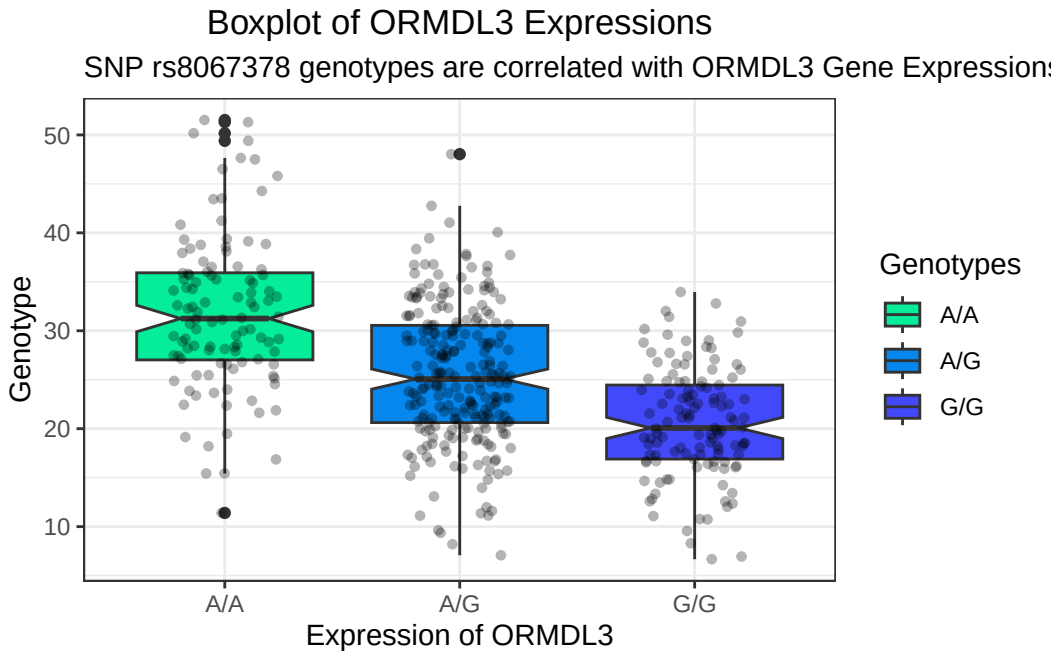
```
   sample              geno                exp
 Length:121         Length:121         Min.   : 6.675
 Class :character   Class :character   1st Qu.:16.903
 Mode  :character   Mode  :character   Median :20.074
                                       Mean   :20.594
                                       3rd Qu.:24.457
                                       Max.   :33.956
```

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)

ggplot(snpfile) +
  aes(geno, exp, fill= geno)+
  ylab("Genotype") + xlab("Expression of ORMDL3") +
  scale_fill_manual(name= "Genotypes", values= c("#05ED99", "#0587ED", "#4149FB"))+
  ggtitle("Boxplot of ORMDL3 Expressions",
```

```
        subtitle = "SNP rs8067378 genotypes are correlated with ORMDL3 Gene Expressions")+
geom_boxplot(notch = T) +
geom_jitter(size=1.2, fill="darkgrey", alpha=0.3, width = 0.23) +
theme_bw() +
ggeasy::easy_center_title()
```



According to this boxplot, the median expression levels of the ORMDL3 gene is quite different between the three SNP genotypes. ORMDL3 is most expressed when the individual is homologous A/A in their rs8067378 SNP. On the other hand, individuals with homologous G/G has the lowest expression of ORMDL3 gene.