Class 12

Gonzalez A16745338

Section 1. Proportion og G/G in a population

Downloaded a CSV file from Ensemble

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 
```

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

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

Now lets look at a different population. I picked the GBR.

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

head(gbr)
```

```
  Sample..Male.Female.Unknown. Genotype..forward.strand. Population.s. Father
1                  HG00096 (M)                       A|A ALL, EUR, GBR      -
2                  HG00097 (F)                       G|A ALL, EUR, GBR      -
3                  HG00099 (F)                       G|G ALL, EUR, GBR      -
4                  HG00100 (F)                       A|A ALL, EUR, GBR      -
5                  HG00101 (M)                       A|A ALL, EUR, GBR      -
6                  HG00102 (F)                       A|A ALL, EUR, GBR      -
  Mother
1      -
2      -
3      -
4      -
5      -
6      -
```

Find proportion of G G

```r
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 
```

this varient that is associated with childhood asthma is more frequent in the GBR population the the MKL population.

Now letsdig further…

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. Hint: The read.table(),summary() and boxplot() functions will likely be useful here. There is an example R script online to be used ONLY if you are struggling in vein. Note that you can find the medium value from saving the output of the boxplot() function to an R object and examining this object.There is also the medium() and summary() function that you can use to check your understanding.

```r
data <- read.table("rs8067378_ENSG00000172057.6.txt", header = TRUE)

summary(data)
```

```
    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
```

```r
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
```

```r
summary_data <- data %>%
  group_by(geno) %>%
  summarise(
    Sample_Size = n(),
    Median_Expression = median(exp, na.rm = TRUE)
  )

print(summary_data)
```

```
# A tibble: 3 x 3
  geno  Sample_Size Median_Expression
  <chr>       <int>             <dbl>
1 A/A           108              31.2
2 A/G           233              25.1
3 G/G           121              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 ORMDL3? Hint: An example boxplot is provided overleaf – yours does not need to be as polished as this one.

```
data <- read.table("rs8067378_ENSG00000172057.6.txt", header = TRUE, sep = " ", na.strings =
str(data)
```

```
'data.frame':   462 obs. of  3 variables:
 $ sample: chr  "HG00367" "NA20768" "HG00361" "HG00135" ...
 $ geno  : chr  "A/G" "A/G" "A/A" "A/A" ...
 $ exp   : num  29 20.2 31.3 34.1 18.3 ...
```

```
head(data)
```

```
   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
```

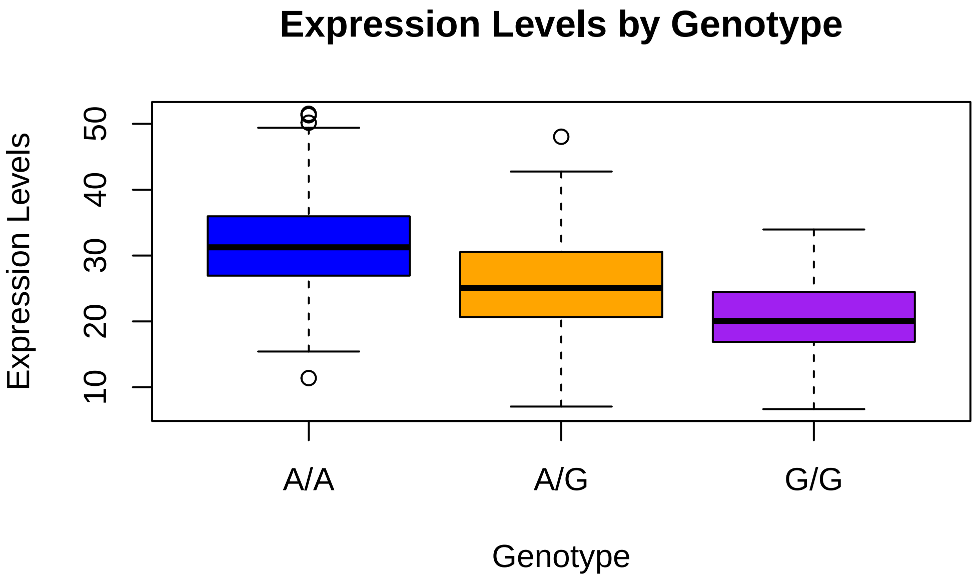
```
summary(data)
```

```
    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  
```

```r
colnames(data) <- c("sample", "geno", "exp")
```

```r
# Filter out rows with NA
data_clean <- data[!is.na(data$exp) & is.finite(data$exp), ]
```

```r
# boxplot
boxplot(exp ~ geno, data = data_clean,
        main = "Expression Levels by Genotype",
        xlab = "Genotype", ylab = "Expression Levels",
        col = c("blue", "orange", "purple"))
```


Expression Levels by Genotype
Expression Levels
A/A
A/G
G/G
Genotype