Week 12 Lab

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Section 1. Proportion og G/G in a population

Downloaded a csv file from Ensemble < https://useast.ensembl.org/Homo_sapiens/Variation/Sample?db=core;r=17:39894595-39895595;v=rs8067378;vdb=variation;vf=959672880#373531_tablePanel

Here we read this csv file

Find the proportion

```
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
```

Now let's look at a different population. I picked the GBR.

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

Find proportion of G|G

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

1

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

This variant is associated with childhood asthma is more frequent in the GBR population than the MKL population.

Lets now dig into this further.

Section 4: Population analysis

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.

```r
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
```

```r
nrow(expr)
```
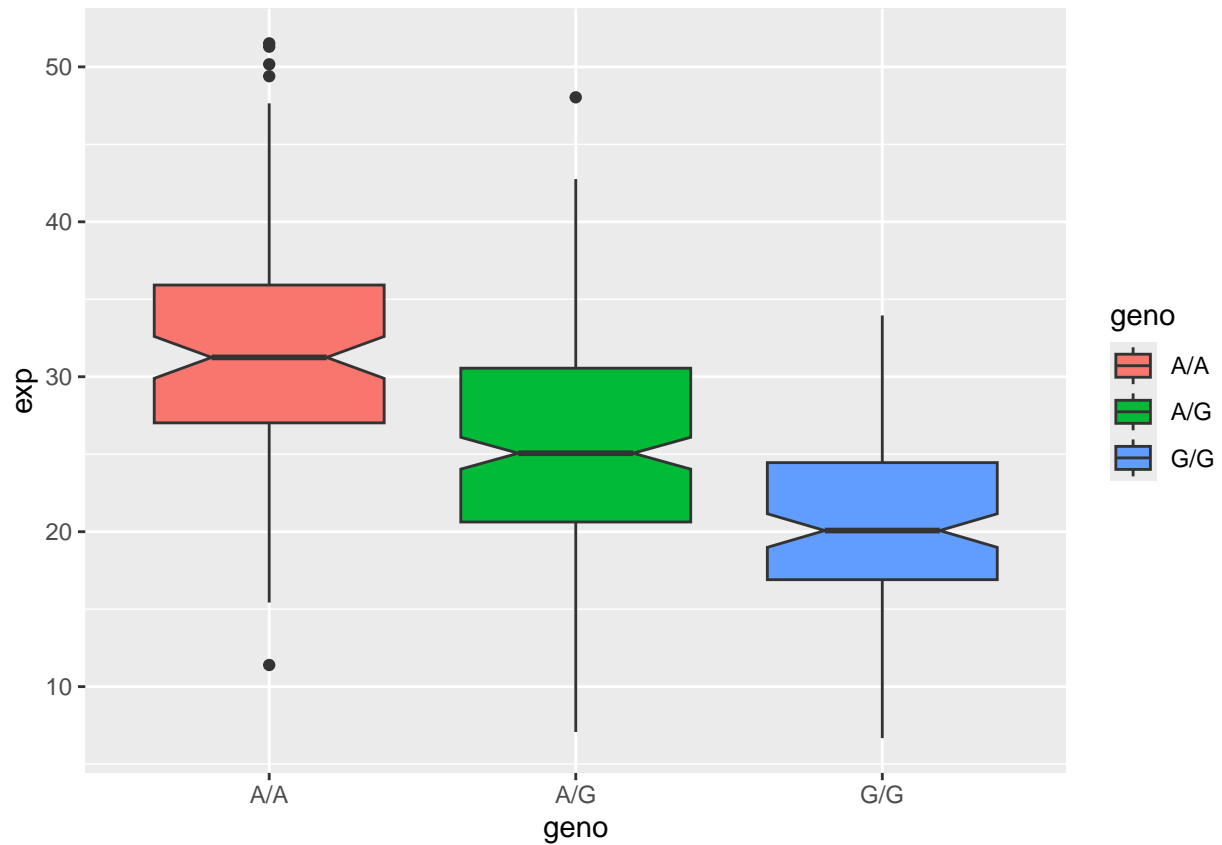
```
## [1] 462
```

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

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

```r
library(ggplot2)
```

Lets make a boxplot

```r
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?

They have different distributions, G/G is significantly different from the A/G and the A/A. The G/G is associated with having a reduced expression of ORMDL3.