Class 12: Ensembl

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Section 1. Propotion og G/G in a population

Downloaded a CSV file from Ensemble < link here

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 let's look at a different population. I pickd 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 the proportion of G|G

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

```
     A|A      A|G      G|A      G|G 
25.27473 18.68132 26.37363 29.67033 
```

This varient that is associated with childhood asthma is more frequent in the GBR population than the MXL population.

Lets now dig into this further.

Section 4: Population Scale Analysis

One smaple 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
```

> 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)
```

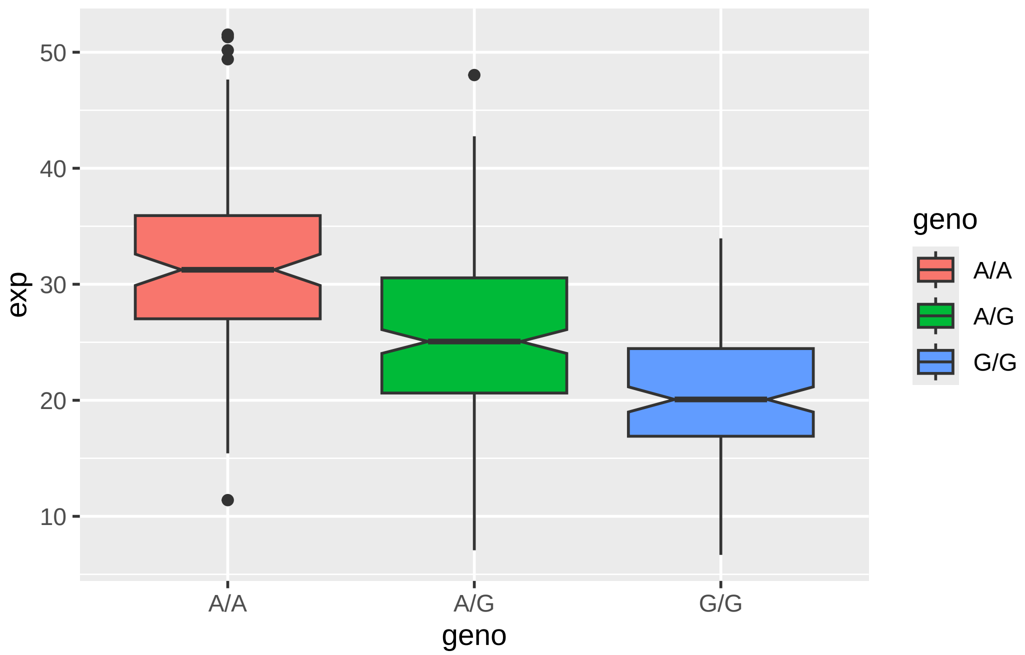
```

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

```
library(ggplot2)
```

Lets make a boxplot

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



```
summary(expr)
```

```
   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  
```

> 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?

based on the boxplot it seems as though the SNP does effect the expression of ORMDL3.