class12

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Section 1. Proportion og G/G in a population

Download a CSV file from Emsemble < https://useast.ensembl.org/Homo_sapiens/Variation/Sample?db=core;r
39898742;v=rs8067378;vdb=variation;vf=959672880#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 
```

```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 picked GBR.

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

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 variant that 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 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. So, you processed about ~230 samples and did the normalization on a genome level. Now, you want to find whether there is any association of the 4 asthma-associated SNPs (rs8067378…) on ORMDL3 expression.

> 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

How many samples do we have?

```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
```

```
nrow(expr)
```

```
[1] 462
```

```
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
```

Sample size for each genotype:

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

```

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

Median expression levels for each genotype:

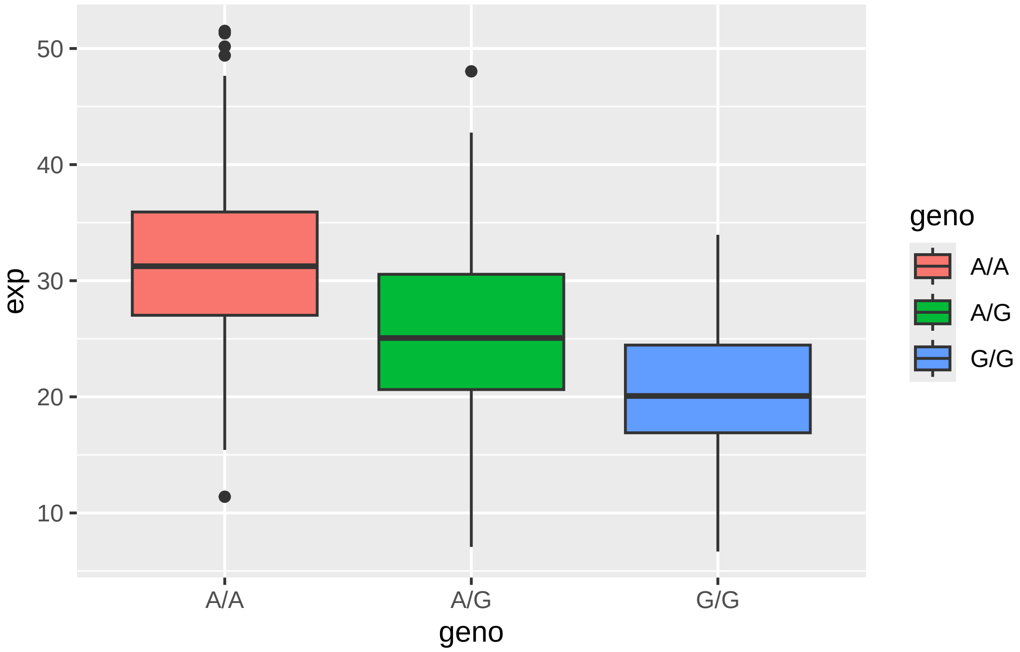
```
round( tapply(expr$exp, expr$geno, median), 2 )
```

```
  A/A   A/G   G/G
31.25 25.06 20.07
```

> 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?

```r
library(ggplot2)

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



G/G has a lower median and overall lower expression of the ORMDL3 gene than A/A. This indicates that the SNP effects the expression of ORMDL3, specifically the G allele in this location is associated with reduced expression of the ORMDL3 gene.