Class 11: Galaxy

Porportion of G/G in a population

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.cs
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.)/ nrow(mxl) *100
```

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

```
gbr <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.cs
```

Find a proportion of G | G

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

```
  A|A   A|G   G|A   G|G
34.38 32.81 18.75 14.06
```

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.

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
```

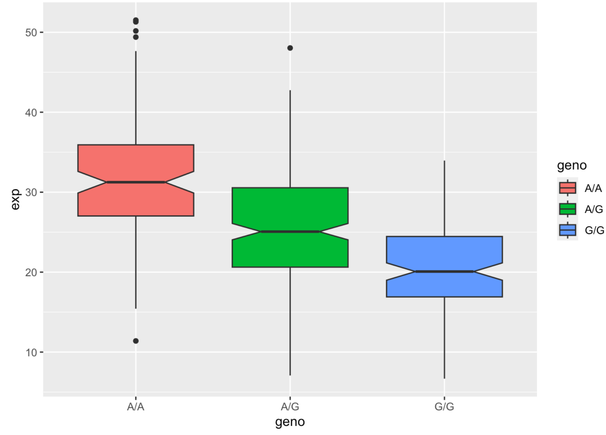
```
table(expr$geno)
```

```
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)
```



> 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?

A/A is the dominant genotype in the population and G/G is less expressed in this population which suggest an enviromental advantage for A/A. It does not seem that the SNP effects the expression of ORMDL3.