Class 12 RNA SeqGalaxy A16246401

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Class 12 RNA SeqGalaxy, Proportion of G/G in a Population

Downloaded CSV file from Ensemble

We are going to read the 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
```

```
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 we are going to look at a GBR population with 91 individuals.

```
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      -
```

```
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 variant that is associated with childhood asthma is more frequent in the GBR population that the MXL population.

Now let's dig into this further.

Section 4 (Homework)

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
```

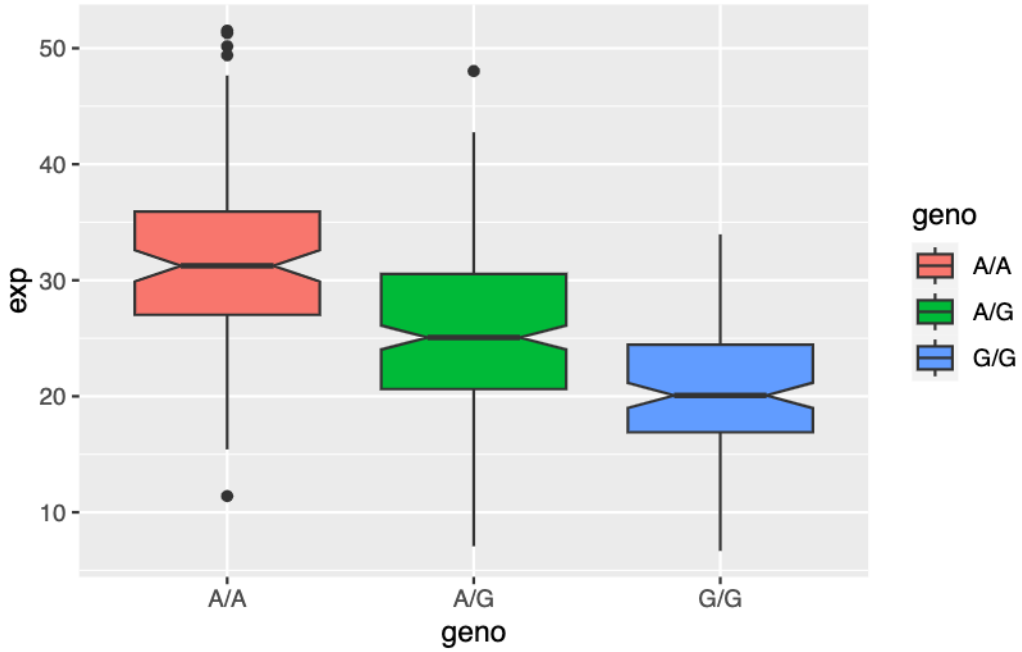
```
table(expr$geno)
```

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

```
library(ggplot2)
```

Let's make a boxplot

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



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.

The boxplot shows the median expression for each of the genotypes. For the allele A/A the median expression is around 31, while A/G is 25, and G/G is 20.

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?

There is a statistically significant difference in expression between the two homozygous genotypes of A/A and G/G. This means that the SNP does effet the expression of ORMDL3.