Class 11: Introduction to Genome Informatics 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
39936599;v=rs8067378;vdb=variation;vf=105535077#373531_tablePanel

Here we read this CSV file

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378 (1).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 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 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 astham is more frequent in the GBR population than the MKL population.

Let’s now dig into this further.

Population Scale 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.

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)
```

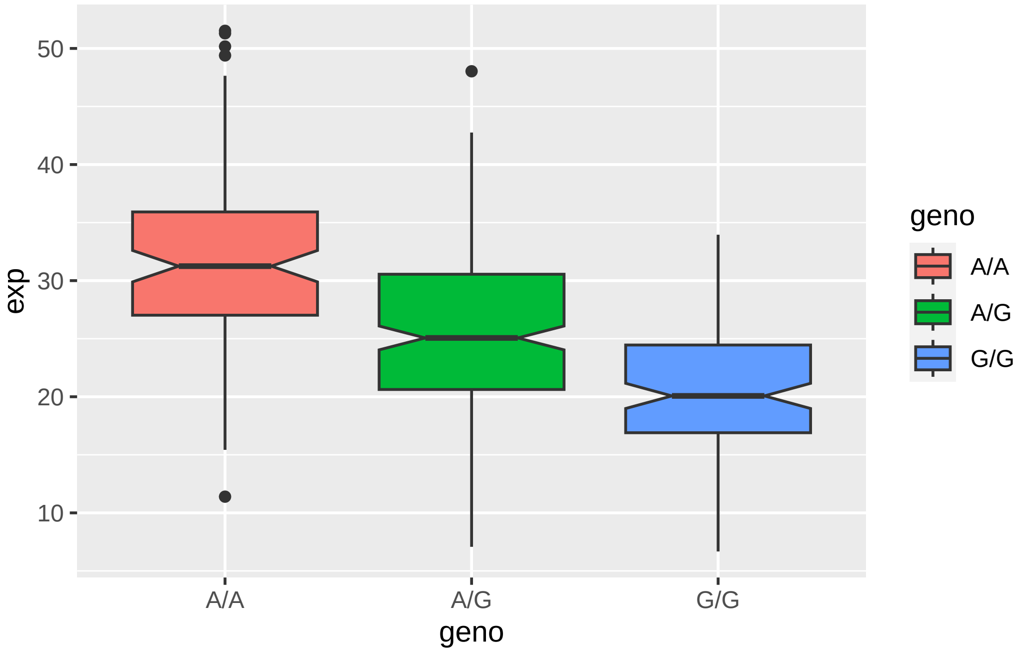
```

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)
```



It looks like the SNP between the G and A affects expression levels. A/A median expression ~ 32 A/G median expression = 25 G/G median expression = 20 A/A has more expression than A/G and G/G, so it looks like the A allele is induces more expression.