# Lab11

#Section 1. Proportion og G/G in a population

```
Downloaded csv file from Ensemble https://uswest.ensembl.org/Homo_sapiens/Variation/
Sample?db=core;r=17:39894595-39895595;v=rs8067378;vdb=variation;vf=105535077
Here we read this csv file
  mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")</pre>
  head(mxl)
  Sample..Male.Female.Unknown. Genotype..forward.strand. Population.s. Father
                    NA19648 (F)
                                                        A|A ALL, AMR, MXL
1
2
                                                        G|G ALL, AMR, MXL
                    NA19649 (M)
3
                                                        A|A ALL, AMR, MXL
                   NA19651 (F)
                                                        G|G ALL, AMR, MXL
4
                   NA19652 (M)
5
                   NA19654 (F)
                                                        G|G ALL, AMR, MXL
6
                    NA19655 (M)
                                                        A|G ALL, AMR, MXL
 Mother
1
2
3
4
5
  View(mxl)
  table(mxl$Genotype..forward.strand.)
```

```
A|A A|G G|A G|G
 22 21 12
  table(mxl$Genotype..forward.strand.) / nrow(mxl) *100
    AIA
            AG
                    G|A
                             G|G
34.3750 32.8125 18.7500 14.0625
Let's look at a different population (GBR)
  gbr <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")</pre>
  head(gbr)
  Sample..Male.Female.Unknown. Genotype..forward.strand. Population.s. Father
1
                    HG00096 (M)
                                                       A|A ALL, EUR, GBR
2
                    HG00097 (F)
                                                       G|A ALL, EUR, GBR
3
                    HG00099 (F)
                                                       G|G ALL, EUR, GBR
                                                       A|A ALL, EUR, GBR
4
                    HG00100 (F)
5
                    HG00101 (M)
                                                       A|A ALL, EUR, GBR
6
                    HG00102 (F)
                                                       A|A ALL, EUR, GBR
  Mother
1
2
3
4
5
Find proportion of G|G
  table(gbr$Genotype..forward.strand.)
A|A A|G G|A G|G
 23 17 24 27
```

```
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 MXL population.

## 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  $\sim 230$  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?

```
#download file from our classroom page
#rename to expr
expr <- read.table("rs8067378_ENSG00000172057.6.txt")
#View the first 6 rows of the file named expr
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
```

```
#summary() tells us more information about the data imported
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

#Subsets all the rows that are A/A genotype
expr\$geno == "A/A"

[1] FALSE FALSE TRUE TRUE FALSE TRUE FALSE FALSE FALSE FALSE [13] TRUE FALSE TRUE TRUE FALSE TRUE FALSE FALSE FALSE FALSE TRUE [25] FALSE FALSE TRUE FALSE FA [37] TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE [49] FALSE FALSE FALSE TRUE FALSE TRUE FALSE FALSE FALSE FALSE FALSE [61] FALSE TRUE FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE FALSE [73] FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE TRUE FALSE FALSE [85] FALSE TRUE FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE [109] FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE Г1337 TRUE FALSE [145] FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE TRUE FALSE [157] FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE TRUE FALSE [169] TRUE FALSE FALSE FALSE TRUE FALSE FALSE TRUE FALSE TRUE TRUE [181] FALSE TRUE FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE [193] FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE TRUE FALSE FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE TRUE FALSE [205] [217] TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE TRUE [229] TRUE TRUE FALSE FALSE FALSE TRUE FALSE FALSE FALSE [241] FALSE TRUE TRUE FALSE TRUE TRUE FALSE FALSE FALSE FALSE [253] FALSE TRUE [265] TRUE TRUE FALSE FALSE TRUE TRUE FALSE FALSE FALSE TRUE FALSE [277] FALSE TRUE FALSE F [289] FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE [301] FALSE FALSE FALSE TRUE TRUE TRUE FALSE FALSE FALSE FALSE FALSE [313] FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE [325] FALSE [337] TRUE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE [349] FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE TRUE TRUE [361] FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE TRUE TRUE FALSE [373] FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE [385] FALSE TRUE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE FALSE [397] FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE TRUE FALSE FALSE

[409] TRUE FALSE FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE TRUE [421] FALSE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE TRUE FALSE F

##Subsets in column exp by all the rows that are A/A genotype
expr\$exp[expr\$geno == "A/A"]

[1] 31.32628 34.11169 32.89721 47.64556 30.71355 25.44983 34.24915 35.15014 [9] 24.54684 49.39612 28.20755 28.56199 51.51787 38.10956 31.13741 39.12999 [17] 32.44173 34.03260 36.27151 26.10355 19.48106 38.77623 40.82922 46.50527 [25] 33.07320 25.47283 29.50655 27.48438 35.69719 32.42236 28.91526 19.14544 [33] 22.85572 31.17067 27.87464 27.91580 16.86780 23.66127 28.03403 36.51922 [41] 27.43637 35.63983 38.85161 22.44576 37.06379 35.67637 50.16704 37.94544 [49] 29.15536 28.85309 38.57101 30.89365 39.31537 31.43743 35.26739 22.37043 [57] 28.46943 30.15636 35.88457 32.59723 36.02549 23.86454 28.38114 25.14243 [65] 51.30170 28.14811 29.18390 27.09760 33.89656 33.48253 15.43178 29.45277 [73] 26.80283 43.51943 45.80808 26.55972 24.87330 29.99549 26.68589 39.37193 [81] 38.39523 41.23635 27.13936 36.55643 28.34688 15.42908 47.50579 30.15754 [89] 34.88439 29.29955 32.87767 43.43665 26.56050 21.87746 33.42582 25.38406 [97] 18.20442 21.62336 35.80093 32.26844 34.40756 23.99631 11.39643 35.24632 [105] 23.38376 34.94395 31.92036 44.27738

#Obtains the median of A/A genotype
median(expr\$exp[expr\$geno == "A/A"])

#### [1] 31.24847

expr\$geno == "A/G"

[85] FALSE TRUE TRUE TRUE FALSE TRUE TRUE FALSE FALSE TRUE TRUE FALSE TRUE TRUE FALSE TRUE FALSE FALSE TRUE [97] FALSE TRUE FALSE [109] FALSE FALSE FALSE TRUE FALSE FALSE TRUE FALSE FALSE TRUE [121] FALSE FALSE TRUE TRUE TRUE TRUE FALSE FALSE TRUE TRUE TRUE FALSE [133] FALSE TRUE FALSE TRUE TRUE TRUE TRUE FALSE TRUE TRUE FALSE [145] TRUE TRUE TRUE TRUE TRUE FALSE TRUE TRUE FALSE FALSE FALSE [157] TRUE TRUE FALSE TRUE TRUE FALSE FALSE TRUE TRUE FALSE FALSE TRUE TRUE FALSE FALSE TRUE FALSE FALSE FALSE [169] FALSE FALSE FALSE [181] TRUE FALSE TRUE TRUE TRUE TRUE FALSE FALSE TRUE FALSE TRUE [193] FALSE FALSE TRUE TRUE FALSE FALSE TRUE FALSE FALSE TRUE TRUE [205] FALSE TRUE FALSE TRUE TRUE TRUE FALSE TRUE TRUE FALSE FALSE [217] FALSE FALSE TRUE TRUE TRUE TRUE TRUE FALSE FALSE FALSE FALSE [229] FALSE FALSE TRUE FALSE FALSE TRUE FALSE TRUE TRUE TRUE FALSE FALSE [241] FALSE FALSE FALSE TRUE FALSE FALSE FALSE TRUE FALSE TRUE TRUE [253] FALSE FALSE TRUE TRUE FALSE TRUE FALSE TRUE FALSE TRUE FALSE FALSE [265] FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE TRUE FALSE [277] TRUE FALSE TRUE FALSE TRUE TRUE FALSE TRUE FALSE TRUE FALSE FALSE TRUE TRUE TRUE TRUE FALSE FALSE [289] TRUE FALSE TRUE FALSE FALSE [301] TRUE TRUE TRUE FALSE FALSE FALSE FALSE TRUE TRUE TRUE TRUE [313] TRUE FALSE TRUE FALSE TRUE TRUE FALSE TRUE TRUE FALSE TRUE TRUE TRUE FALSE TRUE FALSE TRUE TRUE [325] TRUE TRUE TRUE TRUE TRUE [337] FALSE TRUE TRUE FALSE FALSE TRUE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE FALSE FALSE TRUE FALSE FALSE [349] TRUE TRUE FALSE TRUE [361] FALSE FALSE TRUE FALSE TRUE TRUE TRUE FALSE FALSE FALSE TRUE [373] FALSE [385] FALSE FALSE TRUE TRUE FALSE FALSE TRUE FALSE TRUE TRUE TRUE TRUE TRUE FALSE TRUE [397] TRUE TRUE TRUE FALSE FALSE TRUE TRUE FALSE [409] FALSE TRUE TRUE FALSE TRUE TRUE TRUE TRUE TRUE TRUE TRUE FALSE [421] FALSE TRUE TRUE FALSE TRUE FALSE TRUE FALSE TRUE TRUE TRUE [433] FALSE TRUE FALSE FALSE TRUE TRUE TRUE TRUE TRUE FALSE FALSE TRUE FALSE TRUE FALSE TRUE TRUE FALSE TRUE TRUE FALSE TRUE FALSE [457] FALSE FALSE FALSE TRUE FALSE TRUE

## expr\$exp[expr\$geno == "A/G"]

```
[1] 28.96038 20.24449 31.48736 33.85374 16.29854 19.73450 13.96175 18.40351 [9] 34.21985 23.32404 23.18606 18.15997 28.03580 30.65270 20.51327 29.72309 [17] 22.50789 31.68959 28.76435 35.85543 37.62403 20.54885 31.04941 18.94583 [25] 32.29483 27.81775 19.89903 48.03410 27.90313 36.47949 20.03116 29.65063 [33] 34.92257 16.71385 16.68151 25.71008 11.60808 19.30953 20.49040 22.37224 [41] 15.20045 20.07459 35.42982 19.10884 24.81087 33.22193 33.40835 22.38996
```

```
[49] 31.31626 9.36055 23.17937 30.63079 19.10420 25.70962 38.34531 19.03976
 [57] 28.81770 42.75662 25.61662 26.48467 28.02486 24.23377 21.03717 33.51752
 [65] 20.73493 33.55650 36.62034 40.06318 33.92744 21.02084 29.95687 25.80393
 [73] 21.49924 34.79575 34.57705 30.03549 16.59638 27.22300 24.35621 22.86793
 [81] 23.19511 33.74015 24.08401 26.39419 32.33359 11.36287 21.16515 17.21277
 [89] 22.73049 25.57669 11.97590 25.35846 33.32411 22.09122 25.19977 30.83577
 [97] 23.63709 19.66773 24.64870 17.03159 22.27101 15.92557 32.54150 22.40203
[105] 27.41638 31.99645 30.05415 20.01602 34.47373 28.75978 29.91249 15.71646
[113] 21.76610 9.62656 28.53965 29.49417 29.55520 19.44178 21.43751 30.40382
[121] 28.50982 21.48847 31.10134 35.99067 26.42877 26.28329 20.72639 21.09140
[129] 23.79292 18.79569 29.54042 18.78700 21.41071 21.29782 33.91853 27.20808
[137] 17.14895 28.72738 37.73840 34.14567 15.36874 29.50350 25.70400 20.07219
[145] 24.53928 17.84487 23.35766 15.71243 19.89034 25.37234 16.12745 24.18529
[153] 21.97051 13.08172 32.00764 30.59653 29.74443 27.88354 11.12451 7.07505
[161] 23.13726 21.09331 8.20002 20.62572 18.24345 11.13478 24.43943 22.24289
[169] 24.67325 41.03924 33.31795 27.98844 31.57994 36.73337 18.45322 23.60431
[177] 24.33489 35.74662 22.75684 34.42403 25.34866 25.06486 36.78028 30.67131
[185] 37.82860 25.44086 29.49548 23.27255 28.69506 26.52036 27.49975 24.66196
[193] 25.81562 22.49429 26.88264 28.98200 34.31875 26.40231 21.67621 27.56805
[201] 16.16277 18.28963 30.55183 20.70871 17.66476 22.79543 24.03419 18.02351
[209] 21.63102 27.54976 14.79717 25.34921 21.34916 21.20080 28.36006 22.43773
[217] 25.56306 19.50634 28.93651 31.79897 30.80067 33.95134 39.43243 23.91465
[225] 20.97560 20.02507 33.44170 17.32813 32.61856 36.77906 16.84776 31.53069
[233] 23.18250
```

```
median(expr$exp[expr$geno == "A/G"])
```

#### [1] 25.06486

```
expr$geno == "G/G"
```

```
[1] FALSE FALSE FALSE FALSE TRUE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE TRUE FALSE FA
```

[109] TRUE TRUE TRUE FALSE FALSE TRUE TRUE FALSE TRUE TRUE TRUE FALSE [121] FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE TRUE [133] FALSE FALSE TRUE FALSE FALSE FALSE TRUE FALSE TRUE FALSE [145] FALSE FALSE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE TRUE [157] FALSE FALSE TRUE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE FALSE [169] FALSE TRUE TRUE TRUE FALSE FALSE TRUE FALSE FALSE TRUE FALSE FALSE [181] FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE [193] TRUE TRUE TRUE FALSE FALSE TRUE FALSE TRUE FALSE FALSE FALSE [205] FALSE FALSE TRUE FALSE F [217] FALSE TRUE FALSE FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE [229] FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE TRUE FALSE [241] TRUE FALSE FALSE FALSE FALSE TRUE FALSE FALSE TRUE FALSE FALSE [253] TRUE TRUE FALSE FALSE FALSE FALSE TRUE FALSE TRUE FALSE FALSE FALSE [265] FALSE FALSE TRUE TRUE FALSE FALSE TRUE TRUE FALSE FALSE FALSE [277] FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE TRUE TRUE [289] FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE TRUE FALSE [301] FALSE FALSE FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE [313] FALSE TRUE FALSE TRUE FALSE FA [325] FALSE [337] FALSE FALSE FALSE TRUE FALSE F [349] FALSE FALSE TRUE FALSE FALSE TRUE TRUE TRUE FALSE FALSE FALSE [361] TRUE TRUE FALSE TRUE FALSE FAL [373] TRUE FALSE TRUE TRUE FALSE TRUE TRUE TRUE FALSE TRUE FALSE [385] TRUE FALSE FALSE FALSE FALSE TRUE FALSE TRUE FALSE FALSE FALSE [397] FALSE [409] FALSE [421] TRUE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE [433] FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE [445] FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE [457] TRUE TRUE FALSE FALSE FALSE

# expr\$exp[expr\$geno == "G/G"]

```
[1] 18.25141 17.67473 18.55622 23.10383 30.94554 21.14387 18.39547 12.02809 [9] 17.44761 29.82254 23.01983 13.42470 22.65437 11.07445 28.35841 28.79371 [17] 27.08956 16.11138 26.61928 30.18323 19.40790 19.52301 26.56808 17.34076 [25] 10.74263 16.66051 29.01720 20.69333 21.15677 18.58691 19.04962 22.81974 [33] 32.01142 21.12823 18.61268 19.37093 31.42162 16.67764 19.08659 21.55001 [41] 8.29591 12.58869 17.34109 28.23642 19.99979 25.55413 24.45672 23.53572 [49] 22.48273 14.66862 33.95602 18.26466 16.06661 17.32504 19.14766 12.57599 [57] 22.28749 17.29261 24.18141 16.07627 14.80495 23.46573 28.97074 27.78837
```

```
[65] 23.92355 9.55902 12.35836 22.53910 21.98118 16.40569 25.21931 24.32857 [73] 19.42882 26.56993 13.34557 16.60507 24.85165 21.56943 23.95528 16.18962 [81] 22.53720 26.04123 6.67482 20.07363 19.76527 18.50772 20.14146 18.07151 [89] 6.94390 22.14277 14.23742 19.85388 27.73467 19.02064 14.49816 26.78940 [97] 20.84709 10.77316 12.82128 16.90256 29.60045 14.81945 17.46326 23.26922 [105] 21.39806 18.06320 15.91528 24.80823 26.04514 18.28089 23.24907 17.91118 [113] 21.09502 24.74366 27.40521 24.85772 23.08482 16.56929 16.69044 25.08880 [121] 32.78519
```

```
median(expr$exp[expr$geno == "G/G"])
```

#### [1] 20.07363

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

##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.

There are 108 samples with the A/A genotype. There are 233 samples with the A/G genotype. There are 121 samples with the G/G genotype.

```
A/A: 31.24847 A/G: 25.06486 G/G: 20.07363

median(expr$exp[expr$geno == "A/A"])

[1] 31.24847

median(expr$exp[expr$geno == "A/G"])

[1] 25.06486

median(expr$exp[expr$geno == "G/G"])

[1] 20.07363

#Function used to determine sample size per genotype. table(expr$geno)
```

#What to know how many sample are in the dataframe? This function will tell us the number nrow(expr)

#### [1] 462

#Specifically tells us how many samples are found in each type:
table(expr\$geno)

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

#Install ggplot2 so we could make a boxplot

```
library(ggplot2)
```

##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?

Genotype A/A had higher levels of expression than genotype G/G. According to the boxplot the SNP does effect the expression of ORMDL3.

##Following code is used to make ggplot

```
#input the dataframe first
#Aes(include x, y data, fill(is to indicate to fill in the box with the color)=color based
#geom_boxplot: function indicates to make a boxplot
ggplot(expr) + aes(x=geno, exp, fill=geno) + geom_boxplot(notch=TRUE)
```

