Class 17

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ENSEMBL/OMIM Calculations

```
library(dplyr)
```

```
Attaching package: 'dplyr'

The following objects are masked from 'package:stats':

    filter, lag

The following objects are masked from 'package:base':

    intersect, setdiff, setequal, union
```

```
read.csv("MXL_genotypes.csv") -> MXL_genotypes
table(MXL_genotypes)
```

```
, , Population.s. = ALL, AMR, MXL, Father = -, Mother = -

                          Genotype..forward.strand.
Sample..Male.Female.Unknown. A|A A|G G|A G|G
                NA19648 (F)   1   0   0   0
                NA19649 (M)   0   0   0   1
                NA19651 (F)   1   0   0   0
                NA19652 (M)   0   0   0   1
                NA19654 (F)   0   0   0   1
                NA19655 (M)   0   1   0   0
                NA19657 (F)   0   1   0   0
```

NA19658	(M)	1	0	0	0
NA19661	(M)	0	1	0	0
NA19663	(F)	1	0	0	0
NA19664	(M)	0	0	1	0
NA19669	(F)	1	0	0	0
NA19670	(M)	1	0	0	0
NA19676	(M)	0	0	0	1
NA19678	(F)	1	0	0	0
NA19679	(M)	0	1	0	0
NA19681	(F)	0	1	0	0
NA19682	(M)	0	1	0	0
NA19684	(F)	0	1	0	0
NA19716	(F)	0	0	1	0
NA19717	(M)	0	1	0	0
NA19719	(F)	0	0	0	1
NA19720	(M)	0	0	0	1
NA19722	(F)	0	0	1	0
NA19723	(M)	0	0	0	1
NA19725	(F)	0	1	0	0
NA19726	(M)	1	0	0	0
NA19728	(F)	1	0	0	0
NA19729	(M)	0	1	0	0
NA19731	(F)	1	0	0	0
NA19732	(M)	0	1	0	0
NA19734	(F)	0	0	1	0
NA19735	(M)	0	0	0	1
NA19740	(F)	1	0	0	0
NA19741	(M)	1	0	0	0
NA19746	(F)	1	0	0	0
NA19747	(M)	0	0	1	0
NA19749	(F)	0	1	0	0
NA19750	(M)	0	1	0	0
NA19752	(F)	0	1	0	0
NA19755	(F)	1	0	0	0
NA19756	(M)	0	0	1	0
NA19758	(F)	0	1	0	0
NA19759	(M)	0	0	1	0
NA19761	(F)	0	0	1	0
NA19762	(M)	1	0	0	0
NA19764	(F)	1	0	0	0
NA19770	(F)	0	1	0	0
NA19771	(M)	1	0	0	0
NA19773	(F)	1	0	0	0

```
NA19774 (M)   0   1   0   0
NA19776 (F)   0   1   0   0
NA19777 (M)   1   0   0   0
NA19779 (F)   0   0   1   0
NA19780 (M)   1   0   0   0
NA19782 (F)   0   0   1   0
NA19783 (M)   0   1   0   0
NA19785 (F)   1   0   0   0
NA19786 (M)   0   0   1   0
NA19788 (F)   0   1   0   0
NA19789 (M)   0   0   0   1
NA19792 (M)   1   0   0   0
NA19794 (F)   0   0   1   0
NA19795 (M)   0   1   0   0
```

```
sum(MXL_genotypes$Genotype..forward.strand.=="G|G") -> homoz_numb
all_numb = count(MXL_genotypes)
homoz_prop <- (homoz_numb/all_numb) * 100
homoz_prop
```

```
       n
1 14.0625
```

DESeq Analysis

```
genereads <- read.table("genereads.txt", header=TRUE, row.names=1)

summary(genereads)
```

```
    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  
```

```
aa <- sum(genereads$geno == "A/A")
ag <- sum(genereads$geno == "A/G")
gg <- sum(genereads$geno == "G/G")
```

Q13 Sample Size

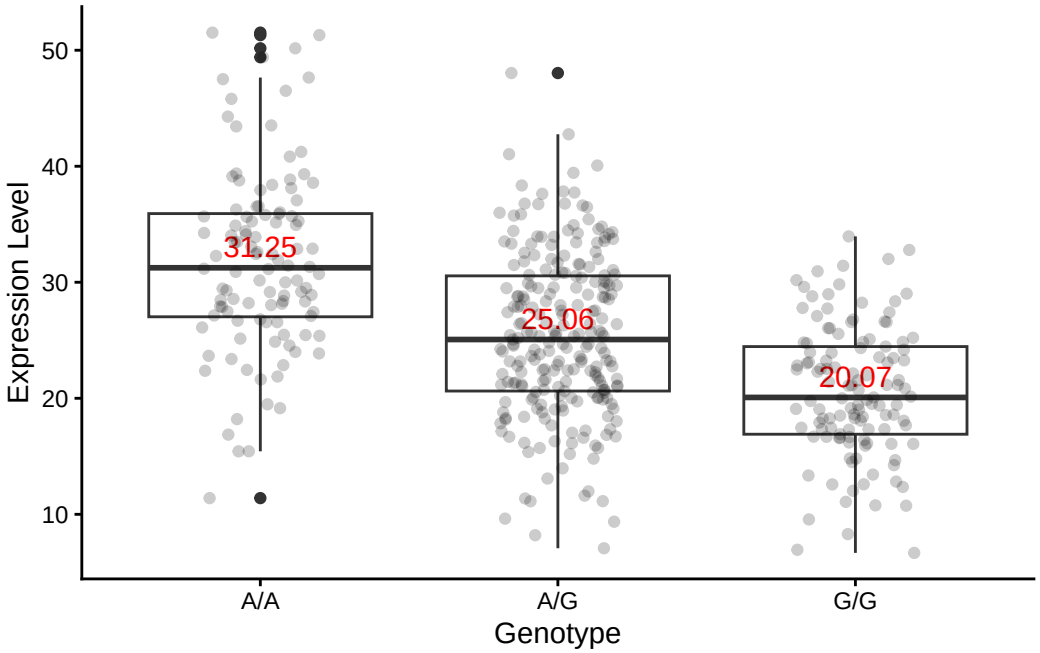
There are 108 individuals with A/A genotype, 233 individuals with A/G genotype, and 121 individuals with G/G genotype.

Median expression provided in boxplot for Q14.

Q14 Box Plot

```
library(ggplot2)
ggplot(genereads) +
  aes(x=geno,y=exp) +
  theme_classic() +
  geom_boxplot() +
   stat_summary(
    fun = median,
    geom = "text",
    aes(label = round(..y.., 2)),
    vjust = -0.5,
    color = "red"
  ) +
  labs(x="Genotype", y="Expression Level") +
  geom_jitter(alpha=0.2, width = 0.2)
```

```
Warning: The dot-dot notation (`..y..`) was deprecated in ggplot2 3.4.0.
i Please use `after_stat(y)` instead.
```



Response to Q14

From the boxplot, I can infer that the more copies there are of the SNP allele variant, the lower the expression of ORMDL3. The allele inhibits proper expression of ORMDL3.