Class 11: Intro to Genome Informatics

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Table of contents

Q5. What proportion of the Mexican Ancestry in Los Angeles sample population (MXL) are homozygous for the asthma associated SNP (G|G)?

14.1%

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
```

```
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
```

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.

- A/A: Sample Size 108, Median Expression 31.2

- A/G: Sample Size 233, Median Expression 25.1

- G/G: Sample Size 121, Median Expression 20.1

```
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(dplyr)
expr.aa <- expr %>% filter(geno=="A/A")
expr.ag <- expr %>% filter(geno=="A/G")
expr.gg <- expr %>% filter(geno=="G/G")
```

```
median(expr.aa$exp)
```

```
[1] 31.24847
```

```
median(expr.ag$exp)
```

[1] 25.06486

```
median(expr.gg$exp)
```

[1] 20.07363

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?

G/G has a lower expression value relative to A/A, and A/G has an intermediate expression value; it seems that G reduces the expression of ORMDL3. From this observation, I would infer that the SNP does affect the expression of ORMDL3.

```
library(ggplot2)
ggplot(expr) +
  aes(x=geno,y=exp,fill=geno) +
  geom_boxplot(show.legend=F, notch=T) +
  geom_jitter(show.legend=F, alpha=0.3) +
  labs(x="Genotype",y="Expression")
```