Class 11

Kalisa Kang (PID A16741690)

Section 1. Proportion of G/G in a population

Downloaded a CSV file from Ensemble < https://useast.ensembl.org/Homo_sapiens/Variation/Sample?db=core
40090102;v=rs8067378;vdb=variation;vf=959672880#373531_tablePanel >

Here we read this 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 let's look at a different population. I picked the GBR (Great Britain) population.

```
gbr <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378 (1).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      -
```

```
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      -
7                  HG00103 (M)                         A|G ALL, EUR, GBR      -
8                  HG00105 (M)                         A|A ALL, EUR, GBR      -
9                  HG00106 (F)                         G|A ALL, EUR, GBR      -
10                 HG00107 (M)                         G|G ALL, EUR, GBR      -
```

11	HG00108 (M)	A|A ALL, EUR, GBR	-
12	HG00109 (M)	G|G ALL, EUR, GBR	-
13	HG00110 (F)	A|G ALL, EUR, GBR	-
14	HG00111 (F)	A|A ALL, EUR, GBR	-
15	HG00112 (M)	G|G ALL, EUR, GBR	-
16	HG00113 (M)	G|G ALL, EUR, GBR	-
17	HG00114 (M)	G|A ALL, EUR, GBR	-
18	HG00115 (M)	A|G ALL, EUR, GBR	-
19	HG00116 (M)	G|G ALL, EUR, GBR	-
20	HG00117 (M)	A|A ALL, EUR, GBR	-
21	HG00118 (F)	G|G ALL, EUR, GBR	-
22	HG00119 (M)	G|A ALL, EUR, GBR	-
23	HG00120 (F)	G|G ALL, EUR, GBR	-
24	HG00121 (F)	A|G ALL, EUR, GBR	-
25	HG00122 (F)	G|G ALL, EUR, GBR	-
26	HG00123 (F)	G|A ALL, EUR, GBR	-
27	HG00125 (F)	A|G ALL, EUR, GBR	-
28	HG00126 (M)	G|G ALL, EUR, GBR	-
29	HG00127 (F)	G|A ALL, EUR, GBR	-
30	HG00128 (F)	A|G ALL, EUR, GBR	-
31	HG00129 (M)	G|G ALL, EUR, GBR	-
32	HG00130 (F)	A|G ALL, EUR, GBR	-
33	HG00131 (M)	G|G ALL, EUR, GBR	-
34	HG00132 (F)	A|A ALL, EUR, GBR	-
35	HG00133 (F)	G|A ALL, EUR, GBR	-
36	HG00136 (M)	G|G ALL, EUR, GBR	-
37	HG00137 (F)	G|A ALL, EUR, GBR	-
38	HG00138 (M)	A|A ALL, EUR, GBR	-
39	HG00139 (M)	G|G ALL, EUR, GBR	-
40	HG00140 (M)	G|A ALL, EUR, GBR	-
41	HG00141 (M)	G|G ALL, EUR, GBR	-
42	HG00142 (M)	G|G ALL, EUR, GBR	-
43	HG00143 (M)	G|A ALL, EUR, GBR	-
44	HG00145 (M)	A|A ALL, EUR, GBR	-
45	HG00146 (F)	A|A ALL, EUR, GBR	-
46	HG00148 (M)	G|A ALL, EUR, GBR	-
47	HG00149 (M)	G|A ALL, EUR, GBR	-
48	HG00150 (F)	G|A ALL, EUR, GBR	-
49	HG00151 (M)	G|A ALL, EUR, GBR	-
50	HG00154 (F)	G|G ALL, EUR, GBR	-
51	HG00155 (M)	A|G ALL, EUR, GBR	-
52	HG00157 (M)	A|A ALL, EUR, GBR	-
53	HG00158 (F)	A|A ALL, EUR, GBR	-

54	HG00159 (M)	A|A ALL, EUR, GBR	-
55	HG00160 (M)	A|A ALL, EUR, GBR	-
56	HG00231 (F)	A|G ALL, EUR, GBR	-
57	HG00232 (F)	G|G ALL, EUR, GBR	-
58	HG00233 (F)	G|G ALL, EUR, GBR	-
59	HG00234 (M)	G|G ALL, EUR, GBR	-
60	HG00235 (F)	A|A ALL, EUR, GBR	-
61	HG00236 (F)	A|A ALL, EUR, GBR	-
62	HG00237 (F)	A|A ALL, EUR, GBR	-
63	HG00238 (F)	G|G ALL, EUR, GBR	-
64	HG00239 (F)	G|A ALL, EUR, GBR	-
65	HG00240 (F)	G|A ALL, EUR, GBR	-
66	HG00242 (M)	G|A ALL, EUR, GBR	-
67	HG00243 (M)	A|G ALL, EUR, GBR	-
68	HG00244 (M)	G|A ALL, EUR, GBR	-
69	HG00245 (F)	A|G ALL, EUR, GBR	-
70	HG00246 (M)	A|G ALL, EUR, GBR	-
71	HG00250 (F)	G|G ALL, EUR, GBR	-
72	HG00251 (M)	G|A ALL, EUR, GBR	-
73	HG00252 (M)	G|A ALL, EUR, GBR	-
74	HG00253 (F)	A|A ALL, EUR, GBR	-
75	HG00254 (F)	A|G ALL, EUR, GBR	-
76	HG00255 (F)	A|G ALL, EUR, GBR	-
77	HG00256 (M)	A|G ALL, EUR, GBR	-
78	HG00257 (F)	G|G ALL, EUR, GBR	-
79	HG00258 (F)	A|A ALL, EUR, GBR	-
80	HG00259 (F)	G|A ALL, EUR, GBR	-
81	HG00260 (M)	G|G ALL, EUR, GBR	-
82	HG00261 (F)	G|G ALL, EUR, GBR	-
83	HG00262 (F)	A|A ALL, EUR, GBR	-
84	HG00263 (F)	G|A ALL, EUR, GBR	-
85	HG00264 (M)	A|G ALL, EUR, GBR	-
86	HG00265 (M)	G|G ALL, EUR, GBR	-
87	HG01334 (M)	A|G ALL, EUR, GBR	-
88	HG01789 (M)	G|A ALL, EUR, GBR	-
89	HG01790 (F)	G|A ALL, EUR, GBR	-
90	HG01791 (M)	A|A ALL, EUR, GBR	-
91	HG02215 (F)	G|G ALL, EUR, GBR	-

	Mother
1	-
2	-
3	-
4	-

```
5       -
6       -
7       -
8       -
9       -
10      -
11      -
12      -
13      -
14      -
15      -
16      -
17      -
18      -
19      -
20      -
21      -
22      -
23      -
24      -
25      -
26      -
27      -
28      -
29      -
30      -
31      -
32      -
33      -
34      -
35      -
36      -
37      -
38      -
39      -
40      -
41      -
42      -
43      -
44      -
45      -
46      -
47      -
```

```
48      -
49      -
50      -
51      -
52      -
53      -
54      -
55      -
56      -
57      -
58      -
59      -
60      -
61      -
62      -
63      -
64      -
65      -
66      -
67      -
68      -
69      -
70      -
71      -
72      -
73      -
74      -
75      -
76      -
77      -
78      -
79      -
80      -
81      -
82      -
83      -
84      -
85      -
86      -
87      -
88      -
89      -
90      -
```

```
91      -
```

Find proportion of G|G

```
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.

Let's now dig into this further.

Section 4: Population Scale Analysis

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)
```

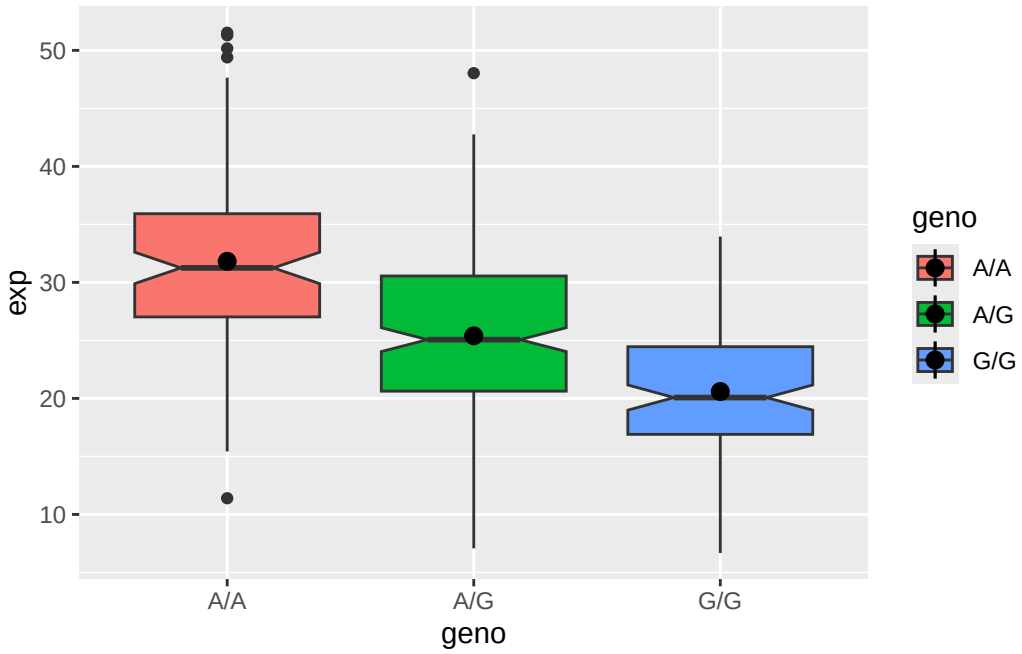
```
A/A A/G G/G
108 233 121
```

```
library(ggplot2)
```

Let's make a boxplot

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

```
No summary function supplied, defaulting to `mean_se()`
```



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.

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

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

The sample size for A|A is 108, A|G is 233, and G|G is 121.

```
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
```

```
medians <- expr %>%
  group_by(geno) %>%
  summarize(median_exp = median(exp))
medians
```

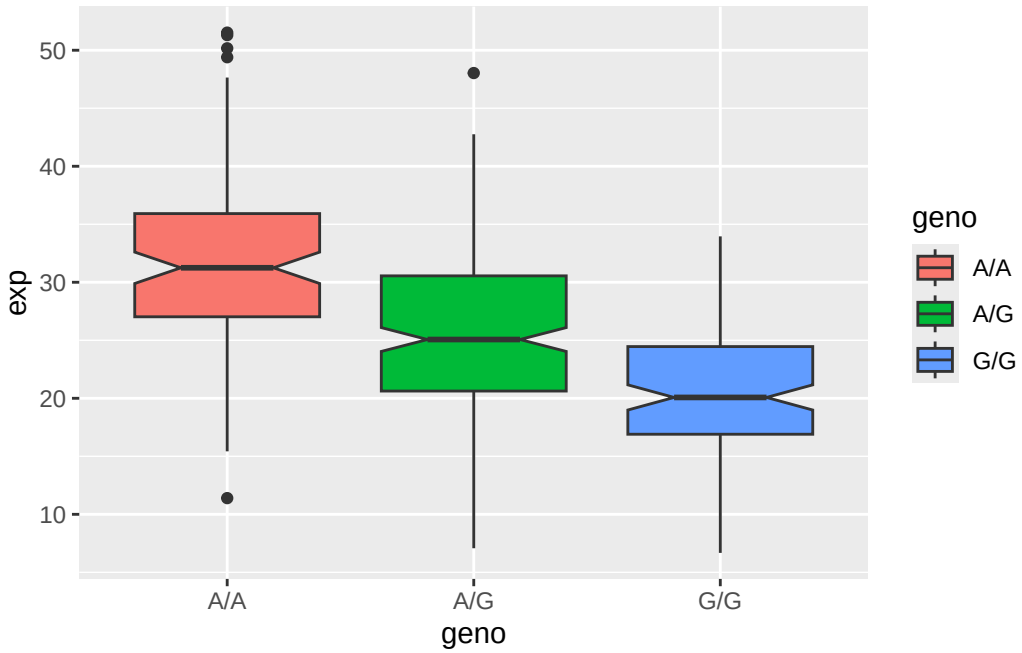
```
# A tibble: 3 x 2
  geno  median_exp
  <chr>      <dbl>
1 A/A         31.2
2 A/G         25.1
3 G/G         20.1
```

The median expression level for A|A is 31.25, A|G is 25.06, and G|G is 20.07.

> 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?

```
library(ggplot2)
```

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



Based on the box plot without any statistical analyses, the expression levels of G|G are lower than that of A|A. The SNP affects expression of ORMDL3.