

Class 12: Genome Informatics

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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.

```
library(tidyverse)
```

```
-- Attaching core tidyverse packages ----- tidyverse 2.0.0 --
v dplyr      1.1.4      v readr      2.1.5
v forcats    1.0.1      v stringr    1.6.0
v ggplot2    4.0.2      v tibble     3.2.1
v lubridate  1.9.4      v tidyr      1.3.1
v purrr      1.0.4
-- Conflicts ----- tidyverse_conflicts() --
x dplyr::filter() masks stats::filter()
x dplyr::lag()     masks stats::lag()
i Use the conflicted package (<http://conflicted.r-lib.org/>) to force all conflicts to become
```

```
samples <- read.table("geno.txt")
```

```
table(samples$geno)
```

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

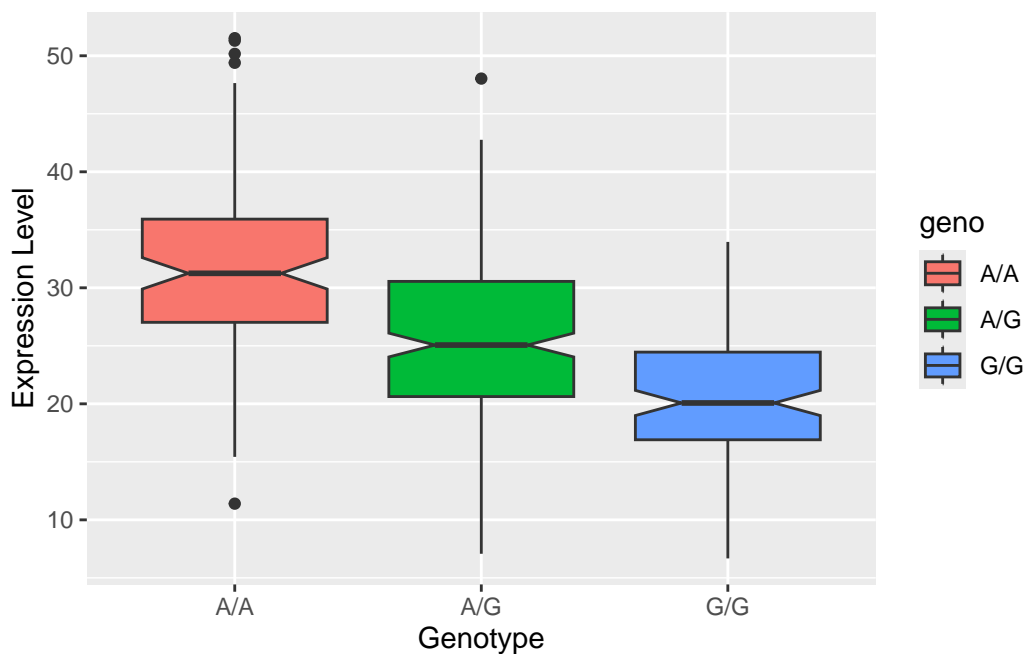
```
samples %>%
  group_by(geno) %>%
  summarize(
    median = median(exp)
  )
```

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

The A/A genotype has a sample size of 108 & median expression level of 31.25, A/G has a sample size of 233 & median expression level of 25.06, and G/G has a sample size of 121 & median expression level of 20.07.

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?

```
ggplot(samples, aes(x = geno, y = exp, fill = geno)) +
  geom_boxplot(notch = TRUE) +
  xlab("Genotype") +
  ylab("Expression Level")
```



Yes, it seems like the SNP decreases the expression level of the G allele of the ORMDL3 gene.