class19.Rmd

Anu Chaparala

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Section 1. Proportion of G/G in Population

Downloaded csv file from https://uswest.ensembl.org/Homo_sapiens/Variation/Sample?db=core;r=17:39825096-39965097;v=rs8067378;vdb=variation;vf=105535077#373531_tablePanel

Read 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)
```

```
##
##      A|A      A|G      G|A      G|G
## 0.343750 0.328125 0.187500 0.140625
```

Section 4: Population Scale Analysis [HOMEWORK] 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 ~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.

```
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
```

> 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.

Median Exp Levels A/A: 32 A/G: 25 G/G: 20

Sample Size A/A: 108 A/G: 233 G/G: 121

```
nrow(expr)
```

```
## [1] 462
```

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

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

```
#summary(plot$data)
```

```
library(ggplot2)
```

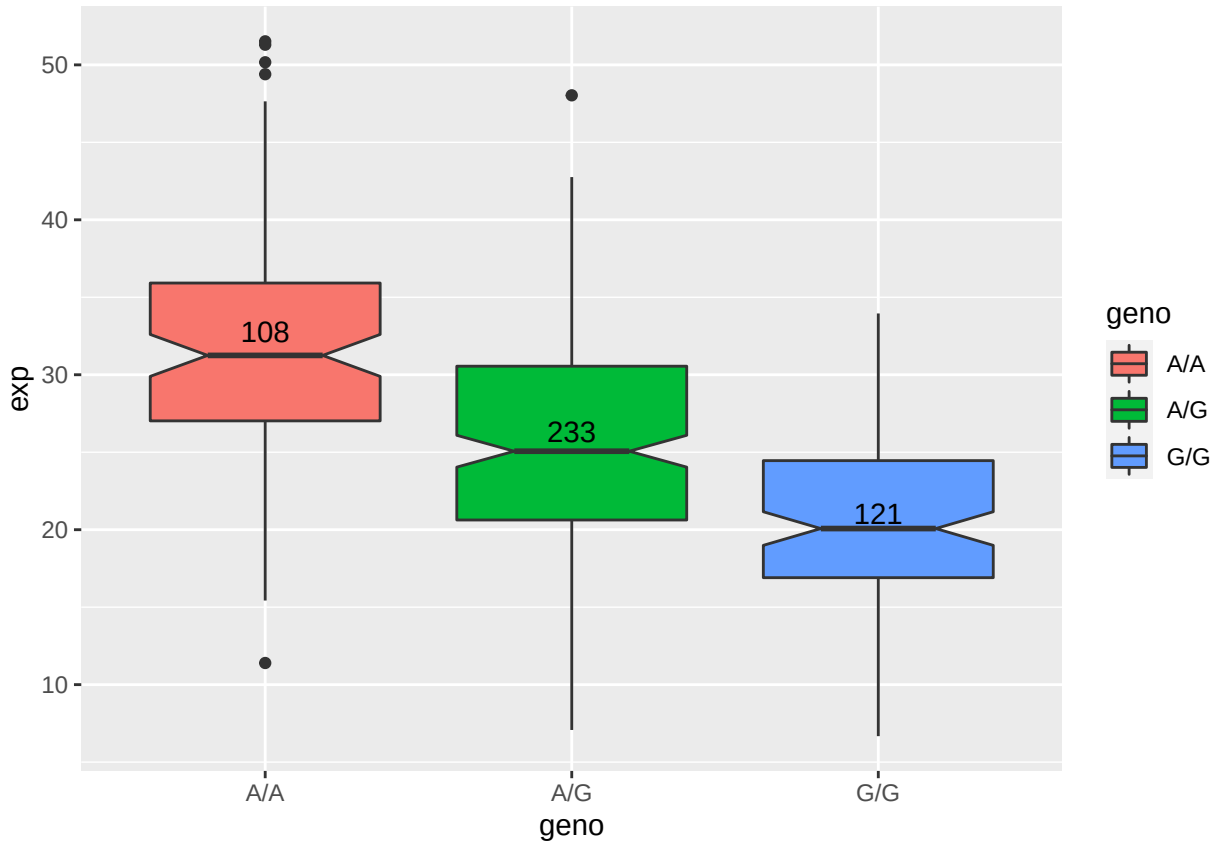
Make box plot

```
give.n <- function(x){
  return(c(y = median(x)*1.05, label = length(x)))
  # experiment with the multiplier to find the perfect position
}

plot <- ggplot(expr, aes(geno, exp, fill=geno)) + geom_boxplot(notch=TRUE)  + stat_summary(fun.data = gi
```

```
## Warning: `fun.y` is deprecated. Use `fun` instead.
```

```
plot
```


50
40
30
20
10
exp
108
233
121
A/A
A/G
G/G
geno
geno
A/A
A/G
G/G