QTL Analysis:
Sugiyama Data

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Description of the data set

In this section, write a short narrative that says something about the data that you are analyzing. You might want to say something about the kinds of mice being bred, the phenotypes contained in the dataset, etc. A good place to look for information is the QTL archives (https://phenome.jax.org/centers/QTLA), and look for your dataset under the name of your author ("List all QTL datasets" is a good place to look!)

Results (text)

Summary of cross

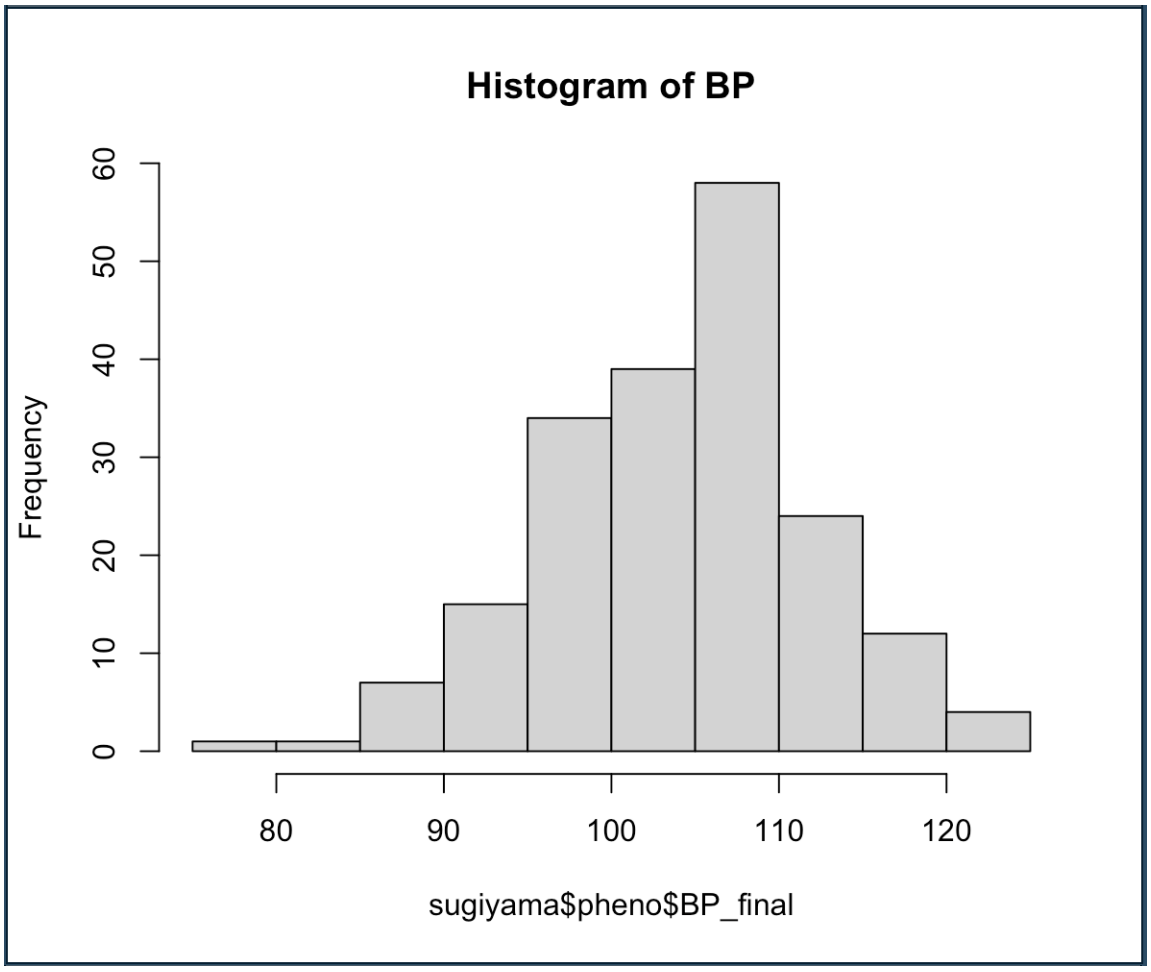
```
No. individuals:    208

No. phenotypes:     7
Percent phenotyped: 100 100 93.8 94.2 99.5 99.5 99.5

No. chromosomes:    20
    Autosomes:      1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19
    X chr:          X

Total markers:      97
No. markers:        5 7 5 5 5 4 8 4 4 5 6 3 3 5 5 4 4 6 5 4
Percent genotyped:  77
Genotypes (%):
     Autosomes:       BB:23.9      BC:50.2      CC:26.0  not CC:0.0    not BB:0.0
   X chromosome:      BY:43.0      CY:57.0
```

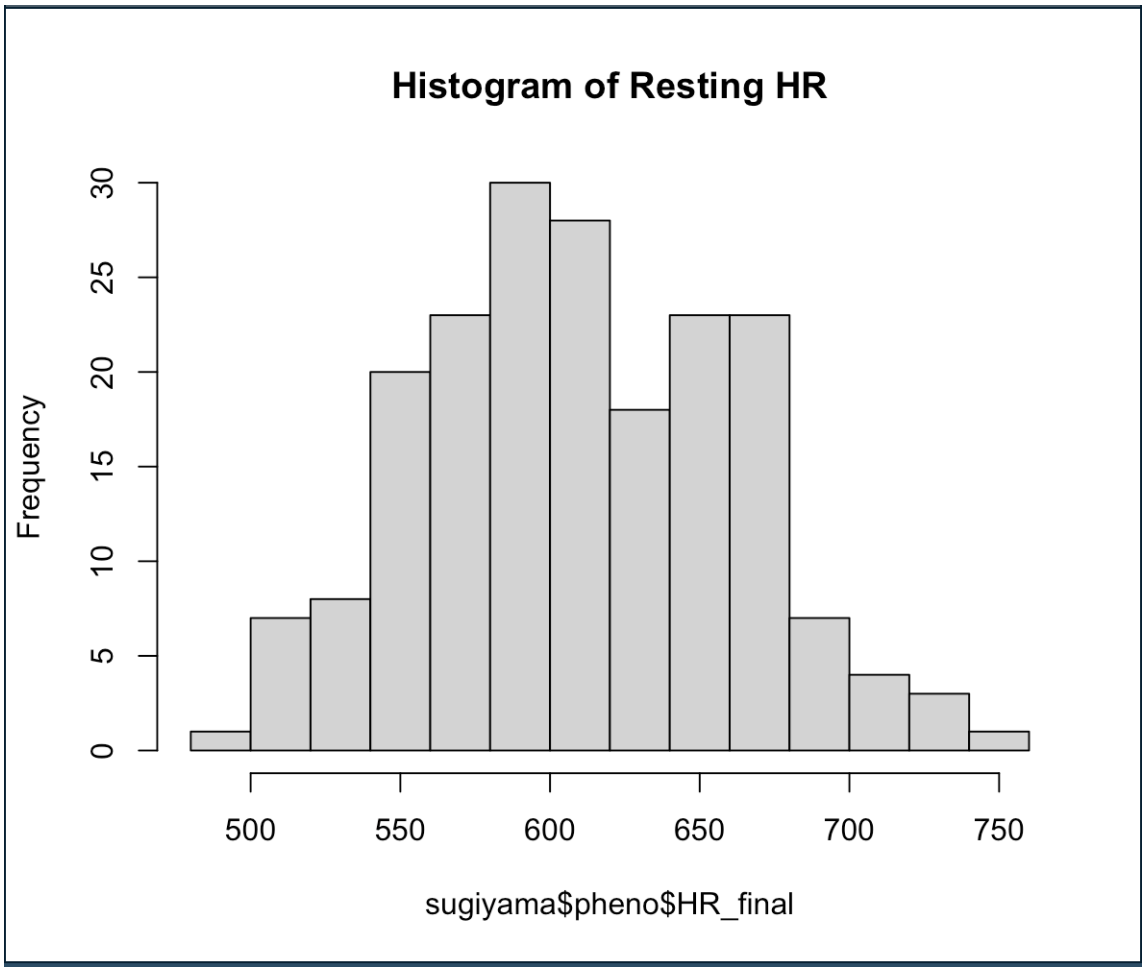
Mainscan results

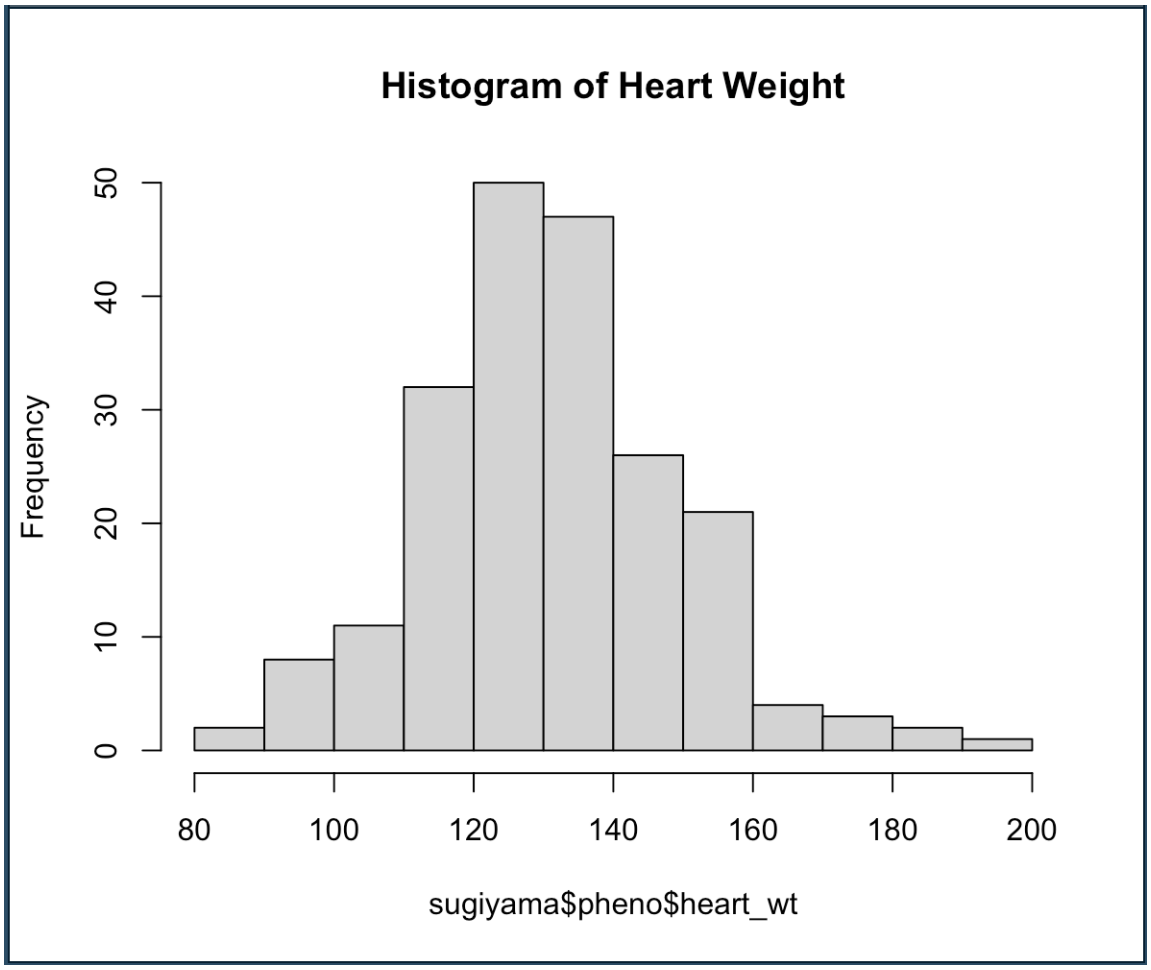
```
        chr  pos  lod
c7.loc46   7 48.7 6.06
c15.loc8  15 12.0 5.17

        chr  pos  lod
c2.loc54   2 59.8 4.15
```

Graphical Results

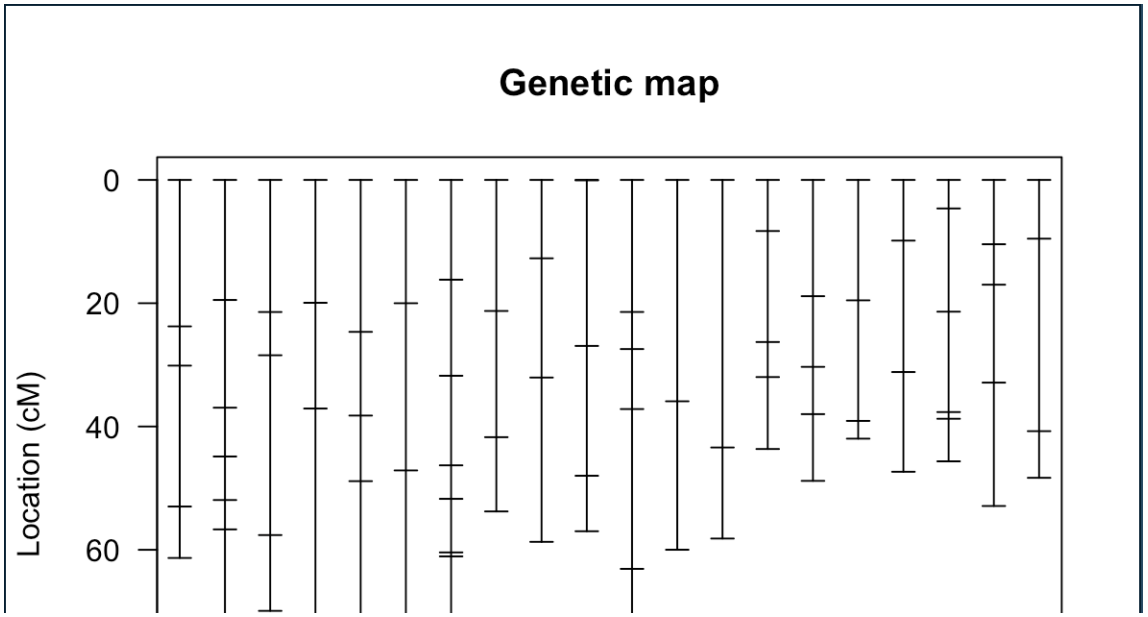
Histogram Plot(s) of phenotype(s)

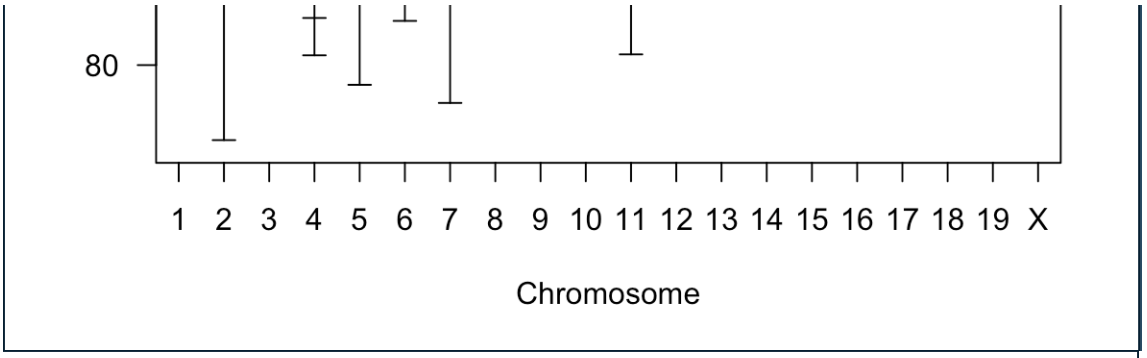

Histogram of BP
Frequency
0
10
20
30
40
50
60
80
90
100
110
120
sugiyama$pheno$BP_final

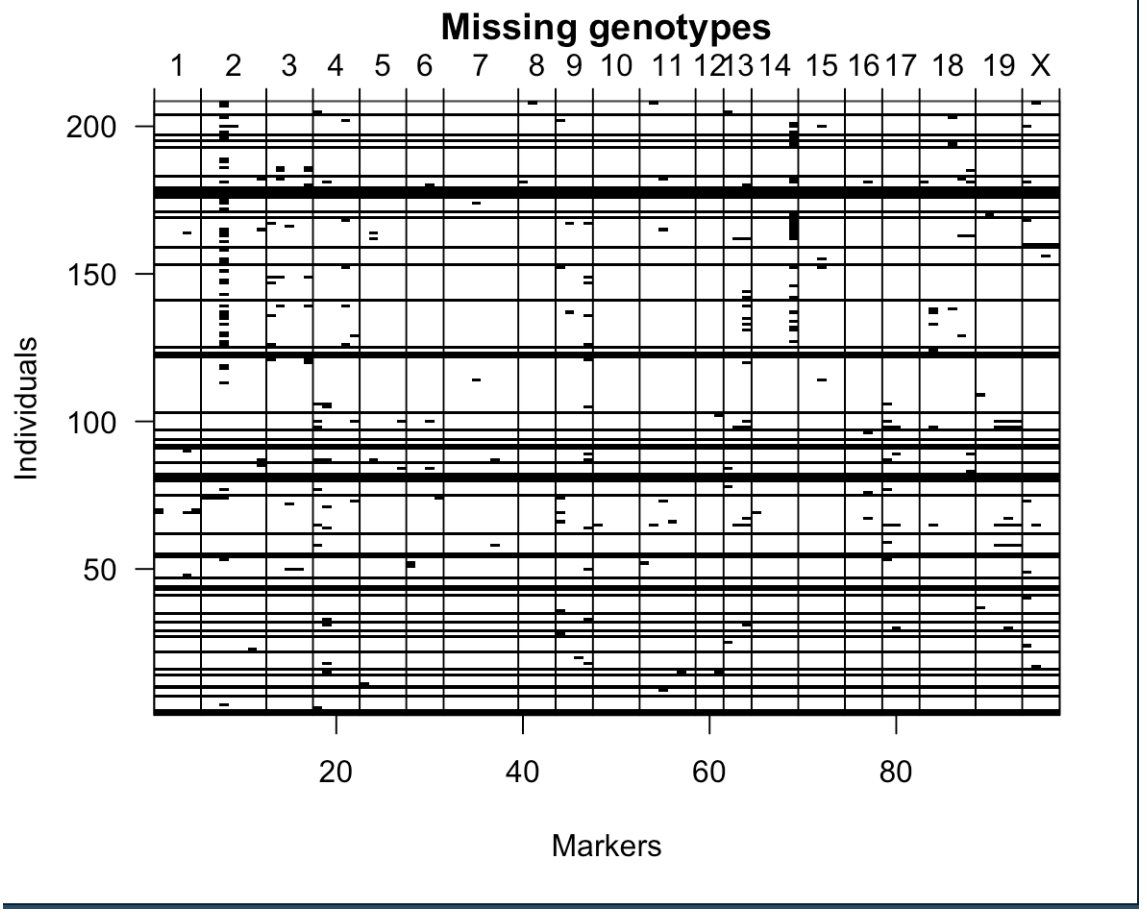

Histogram of Resting HR
Frequency
0
5
10
15
20
25
30
500
550
600
650
700
750
sugiyama$pheno$HR_final


Histogram of Heart Weight
Frequency
0
10
20
30
40
50
80
100
120
140
160
180
200
sugiyama$pheno$heart_wt

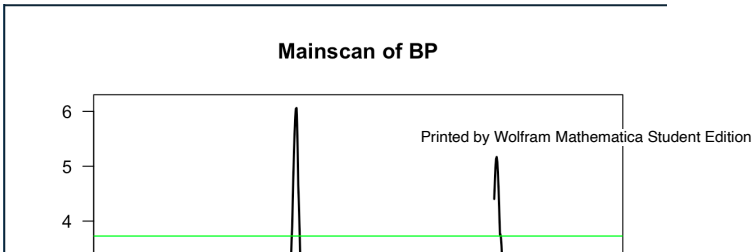
Genetic Map

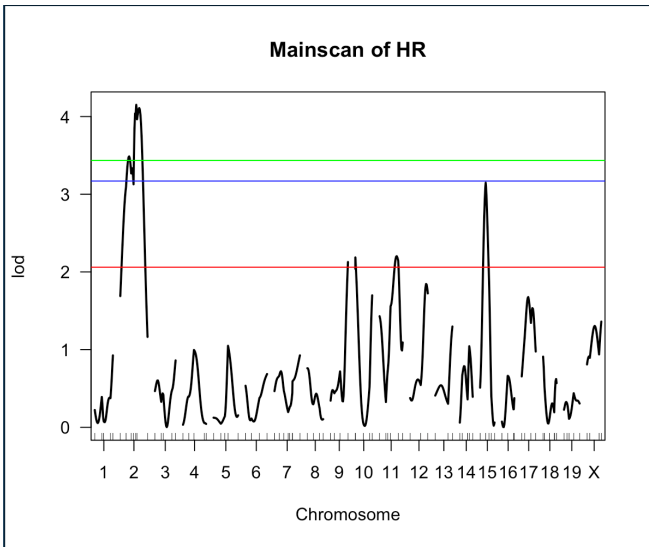

Genetic map
Location (cM)
0
20
40
60

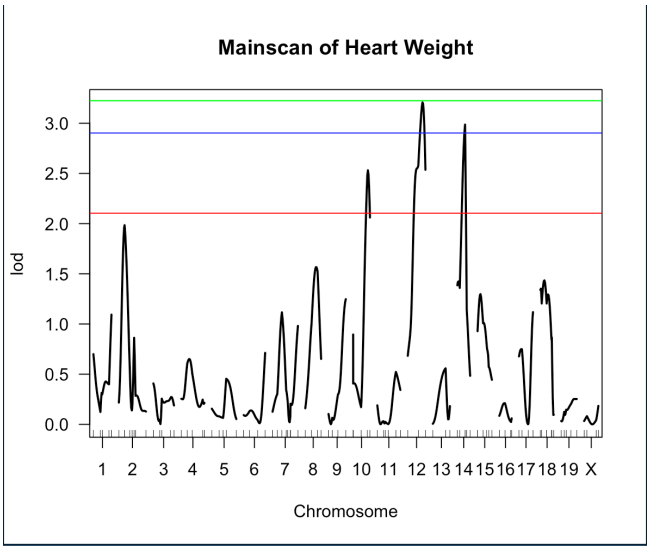




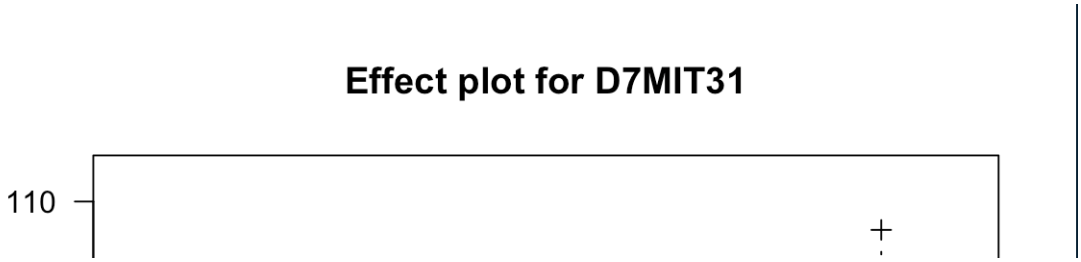
MainScan Plot(s) (with threshold lines at 63%, 10%, and 5%)

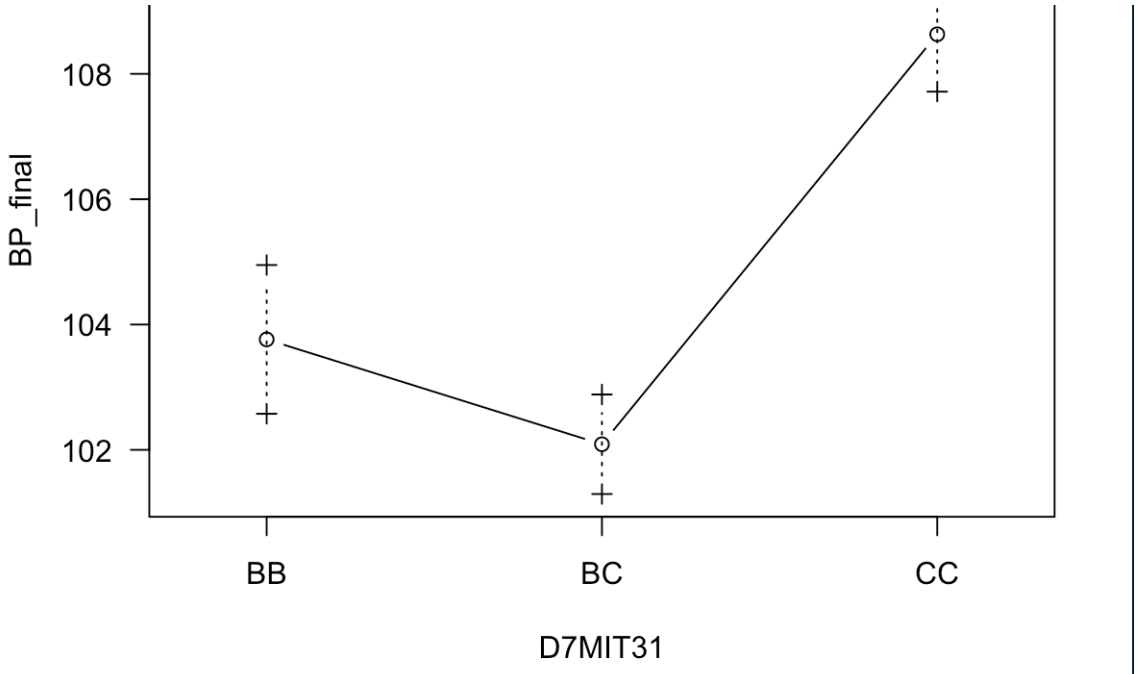


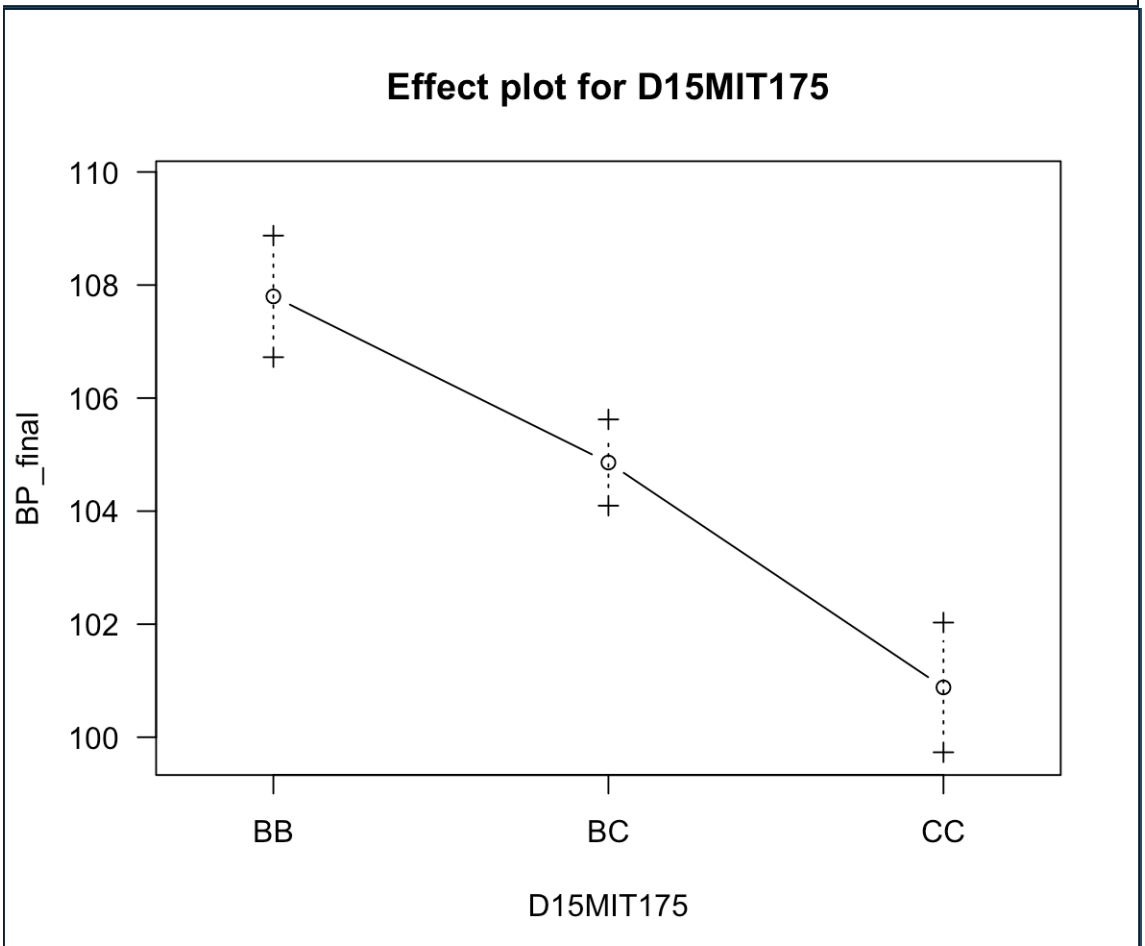


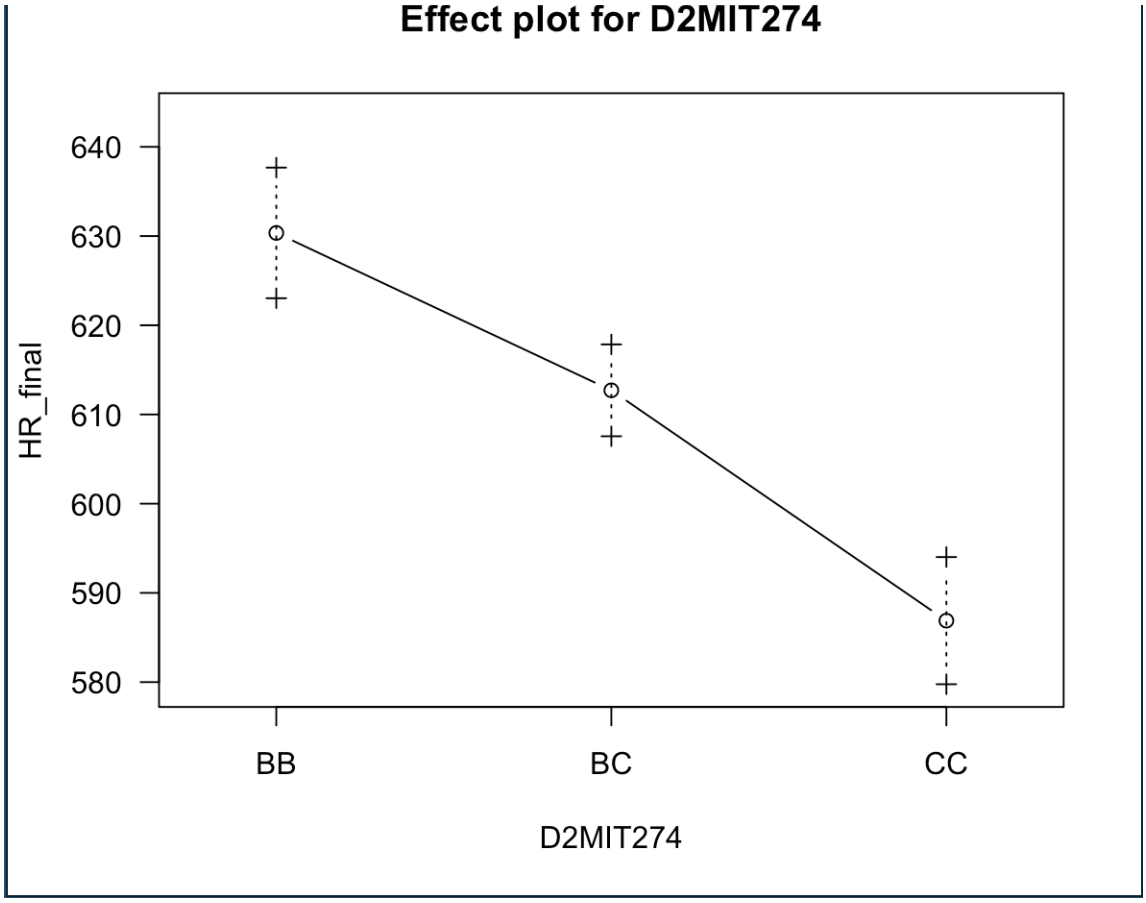


Effect Plot(s) for marker(s) above a LOD threshold of 3




108
106
104
102
BP_final
BB
BC
CC
D7MIT31


Effect plot for D15MIT175
110
108
106
104
102
100
BP_final
BB
BC
CC
D15MIT175



Narrative Analysis

In this section, write a short narrative about what you think you found and the significance of your graphs, especially your QTL scan (mainscan) and your effect plots.

In this lab, we carried out a QTL analysis using a population of BALB/cJ x CBA/CaJ male mice to examine blood pressure, heart rate, and heart weight. Based on the mainscan plot for BP, we see that the genes responsible for this phenotype are likely found on chromosomes 7 and 15. This is corroborated by the two effect plots for BP which display genetic markers on chromosome 7 and 15. The effect plot for D7MIT31 is majority CC, minority BC, and medium BB, while the effect plot for D15MIT175 is majority BB, medium BC, and minority CC. The mainscan plot for HR shows that the genes responsible for this phenotype are likely found on chromosomes 2 and 15. However, the lod score was only significant in chromosome 2, as shown by the effect plot for D2MIT274, which had a similar pattern to the effect plot for D15MIT175. Finally, the mainscan plot for Heart Weight shows that the genes responsible are likely found on chromosomes 2, 10, 12, or 14. However, none of these lod scores were significant, which is why no effect plots were generated.

R/qtl Code

Copy and paste your R/qtl code from R or RStudio here!

```
# R script for analyzing QTL Sugiyama data
# Srijan Oduru
# sugiyamashort.qtl script
# October 10 2021
#
# clean things up
rm(list=ls())
setwd("~/Desktop/R Folder")
#
# load the QTL library
# NOTE!  I first had to INSTALL the library using: install.packages("qtl")
# Now I can use the package qtl
library(qtl)
#
# Load the data!
sugiyama <- read.cross("csv", file="sugiyamashort.csv", genotypes=c("C", "H", "B"),
            na.strings="-", alleles=c("B", "C"))
# Sometimes the genetic markers are too close. Jittermap will move them apart slightly so my results
are better.
jittermap(sugiyama)
#  A summary of the cross gives me some basic data.  Nice!
summary(sugiyama)
# I need to see what phenotypes are in the dataset.  names does that for me
names(sugiyama)
# take a look at my data, make sure it's pretty clean
#  I should NOT see any really big red spots, especially in the bottom right corner under the diagonal
sugiyama <- est.rf(sugiyama)
plotRF(sugiyama)
# It's nice to see my genetic map -- all of the horizontal lines are genetic markers that have been
inserted.
plot.map(sugiyama)
#  It's often the case that I have missing data -- plot.missing shows me where it is
plotMissing(sugiyama)
# a histogram of my BP phenotype -- if I get a "normal" distribution (a bell-shaped curve), then I can be
pretty confident of the data
hist(sugiyama$pheno$BP_final, main="Histogram of BP")
```

```
hist(sugiyama$pheno$HR_final, main="Histogram of Resting HR")
hist(sugiyama$pheno$heart_wt, main="Histogram of Heart Weight")
#  I'm lazy....I hate to type "cross$pheno$bp every time I need to study the BP, so I give it a short name.



#
# Now I'm going to generate a mainscan.  First, I calculate what the scan SHOULD look like, so I'm going to calculate a genetic probability map.
sugiyama <- calc.genoprob(sugiyama, step=2.0, off.end=0.0, error.prob=1.0e-4, map.function="haldane",
                stepwidth="fixed")
#
# Run a simulated geno probability calculations
sugiyama <- sim.geno(sugiyama, step=2.0, off.end=0.0, error.prob=1.0e-4, map.function="haldane",
            stepwidth="fixed")
# Perform the mainscan for the QTL
sugiyama.scanBP <- scanone(sugiyama, pheno.col=3, model="normal", method="em")
#  I'm only going to run this for 100 "permulations" -- typically you do 500-1000, but that takes a LONG time.
sugiyama.scanBP.perm <- scanone(sugiyama, pheno.col=3, model="normal", method="em", n.perm=100)

#
# plot the mainscan
plot(sugiyama.scanBP, main="Mainscan of BP")
#  I'm putting threshold likes at 63% confidence, 90% confidence, and 95% confidence.
thresh <- summary(sugiyama.scanBP.perm, alpha=c(0.63, 0.10, 0.05))
abline(h=thresh[1], col="red")
abline(h=thresh[2], col="blue")
abline(h=thresh[3], col="green")

#2nd iteration of mainscan generation and plotting resting heart rate
sugiyama <- calc.genoprob(sugiyama, step=2.0, off.end=0.0, error.prob=1.0e-4, map.function="haldane",
                stepwidth="fixed")
sugiyama <- sim.geno(sugiyama, step=2.0, off.end=0.0, error.prob=1.0e-4, map.function="haldane",
            stepwidth="fixed")

sugiyama.scanHR_final <- scanone(sugiyama, pheno.col=4, model="normal", method="em")
sugiyama.scanHR_final.perm <- scanone(sugiyama, pheno.col=4, model="normal", method="em",
```

```
n.perm=100)

plot(sugiyama.scanHR_final, main="Mainscan of HR")

thresh <- summary(sugiyama.scanHR_final.perm, alpha=c(0.63, 0.10, 0.05))
abline(h=thresh[1], col="red")
abline(h=thresh[2], col="blue")
abline(h=thresh[3], col="green")
#
#3rd iteration of mainscan generation and plotting heart weight
sugiyama <- calc.genoprob(sugiyama, step=2.0, off.end=0.0, error.prob=1.0e-4, map.function="hal-
dane",
                  stepwidth="fixed")
sugiyama <- sim.geno(sugiyama, step=2.0, off.end=0.0, error.prob=1.0e-4, map.function="haldane",
            stepwidth="fixed")

sugiyama.scanheart_wt <- scanone(sugiyama, pheno.col=6, model="normal", method="em")
sugiyama.scanheart_wt.perm <- scanone(sugiyama, pheno.col=6, model="normal", method="em",
n.perm=100)

plot(sugiyama.scanheart_wt, main="Mainscan of Heart Weight")

thresh <- summary(sugiyama.scanheart_wt.perm, alpha=c(0.63, 0.10, 0.05))
abline(h=thresh[1], col="red")
abline(h=thresh[2], col="blue")
abline(h=thresh[3], col="green")
#
#  I'd like to see a text-based output of my scan
summary(sugiyama.scanBP, perm=sugiyama.scanBP.perm, lodcolumn=1, alpha=0.05)
summary(sugiyama.scanHR_final, perm=sugiyama.scanHR_final.perm, lodcolumn=1, alpha=0.05)
summary(sugiyama.scanheart_wt, perm=sugiyama.scanheart_wt.perm, lodcolumn=1, alpha=0.05)
#
# do an effect plot
# once you see an effect plot, you'll understand what it does!
# first effect plot (bp)
mname1 <- find.marker(sugiyama, chr=7, pos=48.7)
effectplot(sugiyama, pheno.col=3, mname1=mname1)
# second effect plot (bp)
mname2 <- find.marker(sugiyama, chr=15, pos=12.0)
effectplot(sugiyama, pheno.col=3, mname1=mname2)
# third effect plot (hr)
```

```
mname3 <- find.marker(sugiyama, chr=2, pos=59.8)
effectplot(sugiyama, pheno.col=4, mname1=mname3)
```

```
# All done
#EOF
```