QTL Analysis:
Sugiyama 2022

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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

F2 intercross

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: CC:23.9 CB:50.2 BB:26.0 not BB:0.0 not CC:0.0
   X chromosome: CY:43.0 BY:57.0

Mainscan results

BLOOD PRESSURE
   chr   pos   lod
2   : 53 Min. : 1.806 Min. :0.000225
7   : 51 1st Qu.:24.840 1st Qu.:0.227641
5   : 46 Median :40.767 Median :0.530073
11 : 45 Mean :41.722 Mean :0.848277
4   : 44 3rd Qu.:56.688 3rd Qu.:1.038739
6   : 40 Max. :97.965 Max. :6.060793
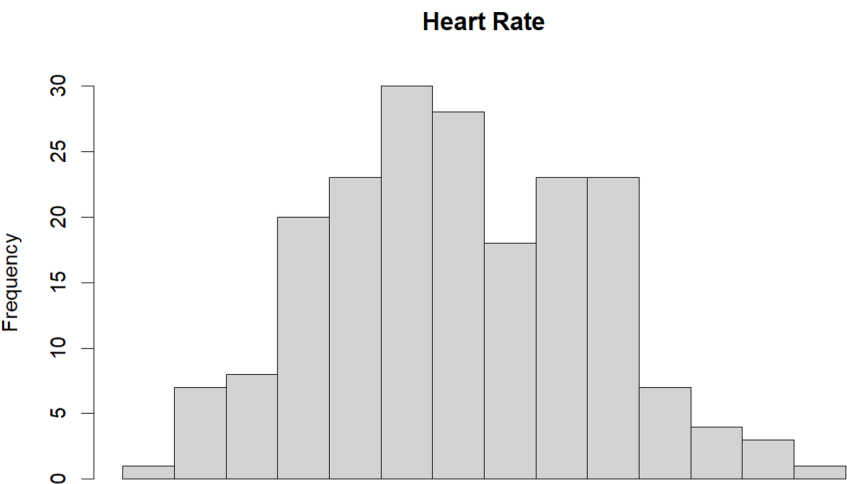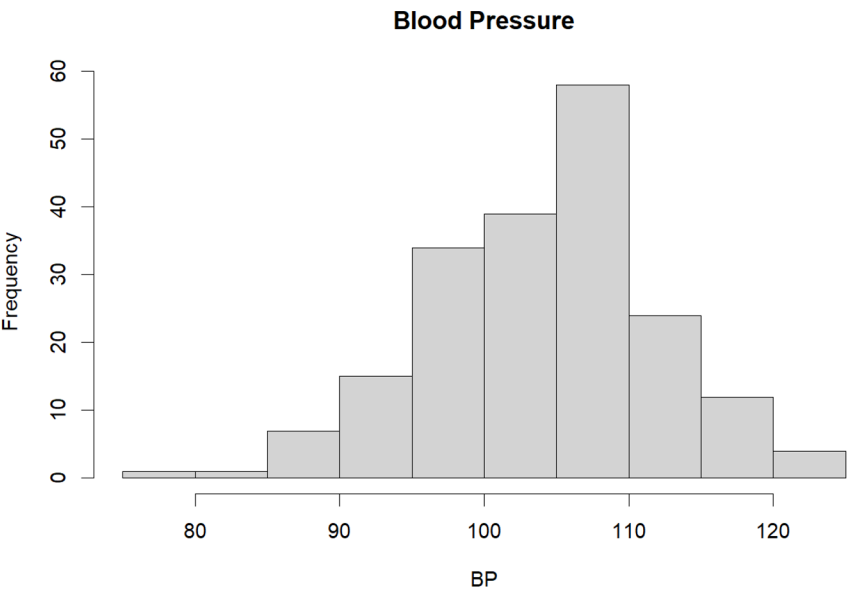HEART RATE
chr   pos   lod
2   : 53 Min. : 1.806 Min. :0.002324
7   : 51 1st Qu.:24.840 1st Qu.:0.298222
5   : 46 Median :40.767 Median :0.519250

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11    : 45   Mean  :41.722   Mean  :0.825131
4     : 44   3rd Qu.:56.688   3rd Qu.:0.994581
6     : 40   Max.  :97.965   Max.  :4.150695
HEART WEIGHT
   chr       pos         lod
2     : 53   Min.  : 1.806   Min.  :0.000034
7     : 51   1st Qu.:24.840   1st Qu.:0.132858
5     : 46   Median :40.767   Median :0.292894
11    : 45   Mean  :41.722   Mean  :0.566419
4     : 44   3rd Qu.:56.688   3rd Qu.:0.757524
6     : 40   Max.  :97.965   Max.  :3.206899
```
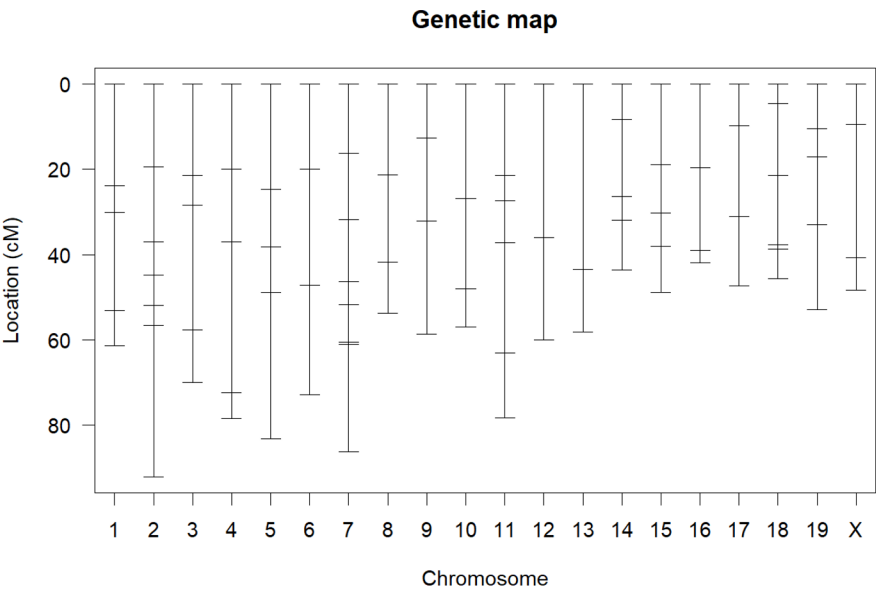
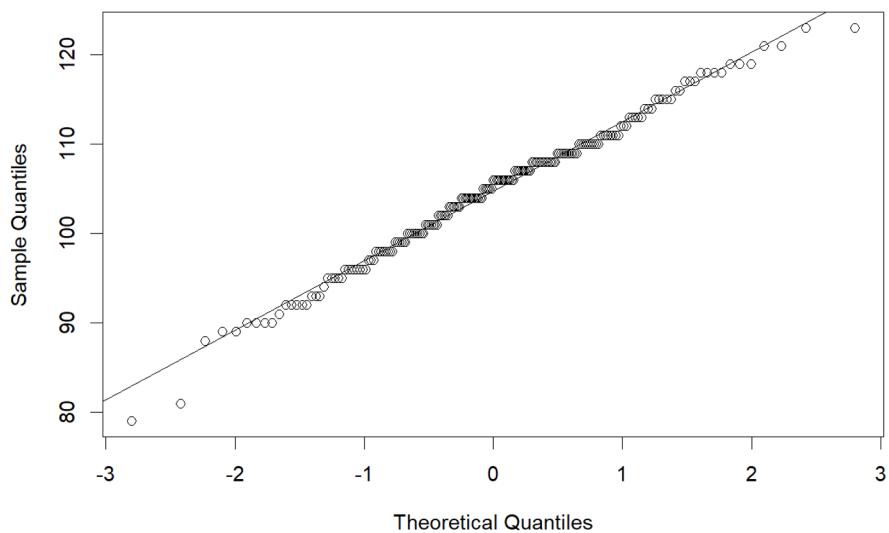# Graphical Results

Histogram Plot(s) of phenotype(s)
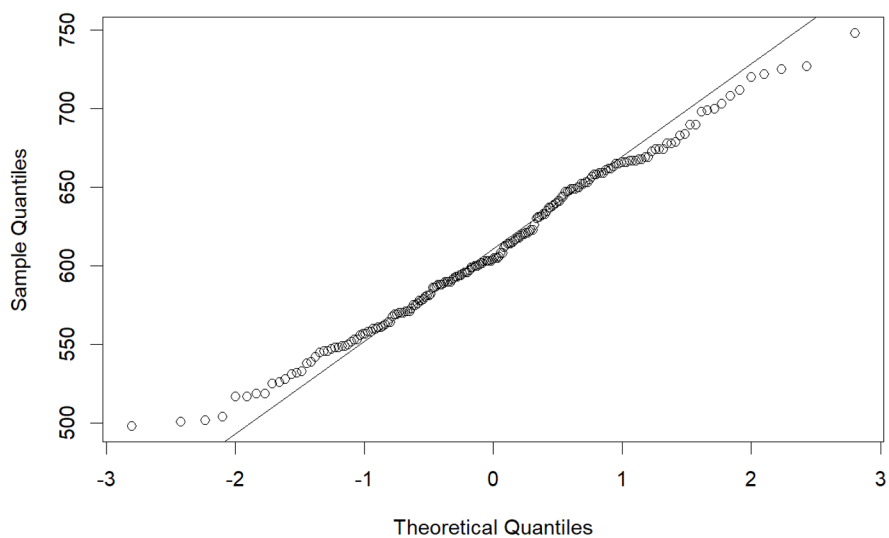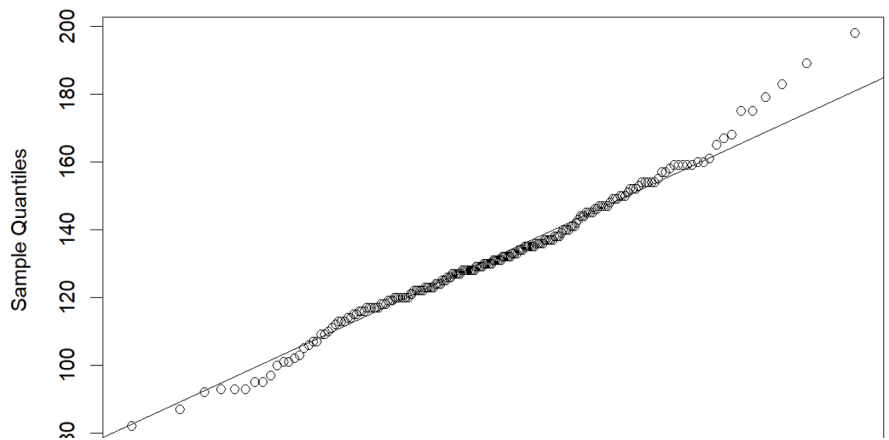

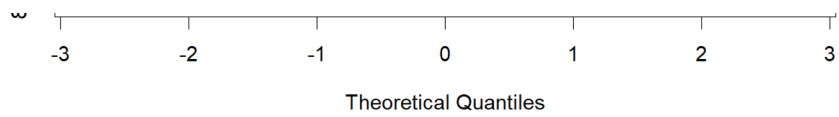**Blood Pressure**

Heart Rate

Heart Weight

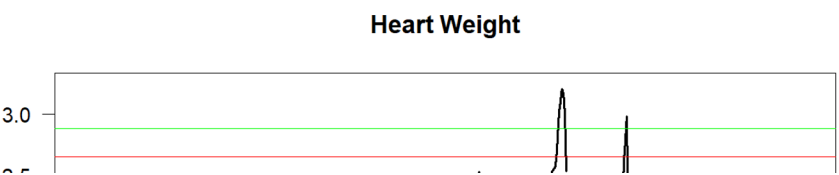**Heart Weight**
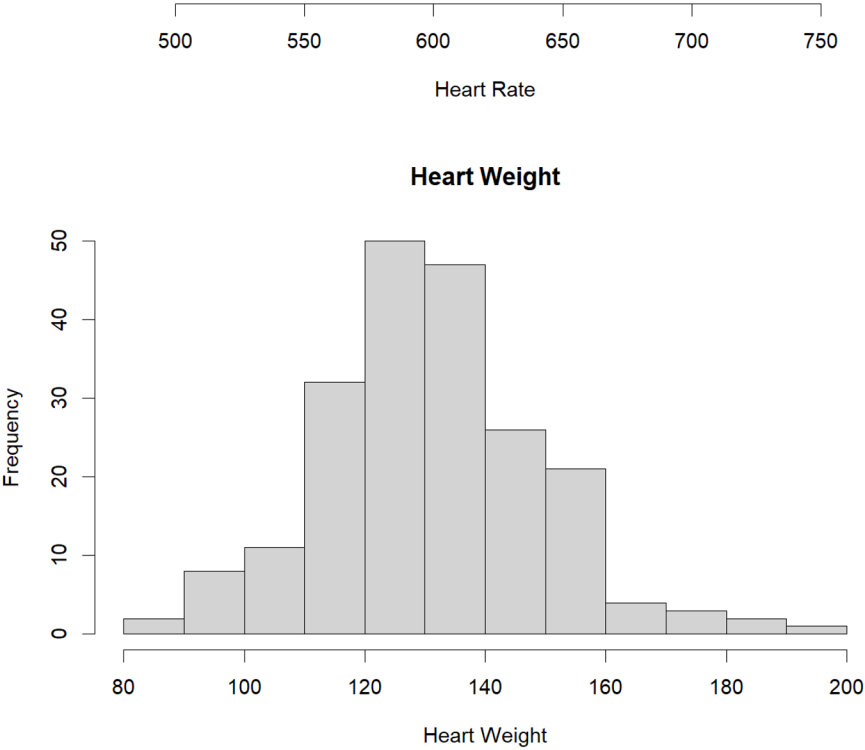
Genetic Map

Genetic map

QQ Plot with regression line

Blood Pressure

Heart Rate

Heart Weight

-3 -2 -1 0 1 2 3

Theoretical Quantiles

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

Blood Pressure

Chromosome

Heart Rate

Chromosome
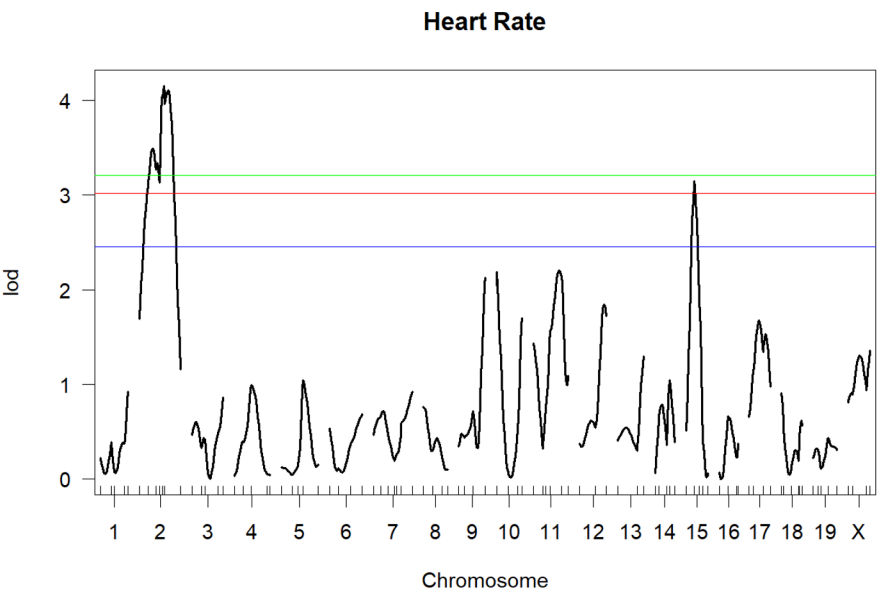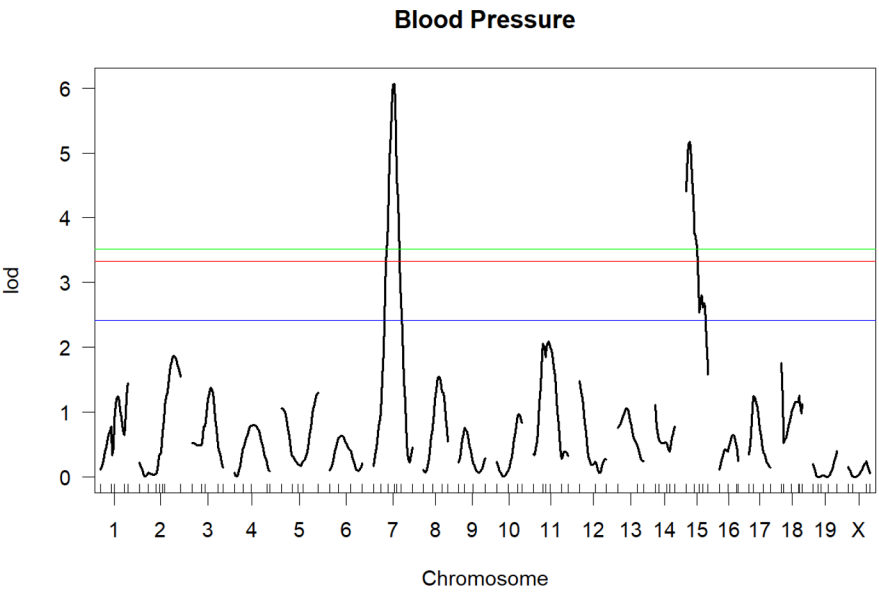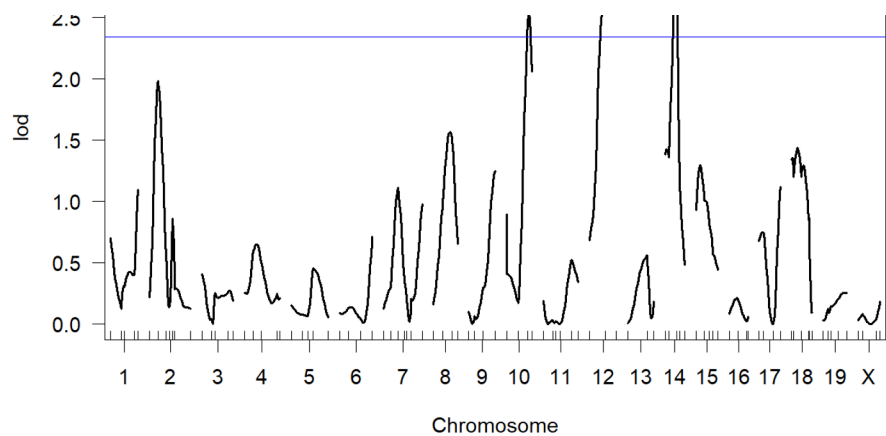
Heart Weight

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

Blood Pressure

Effect plot for D3MIT29

Effect plot for D4MIT181

Heart Rate

Effect plot for D4MIT234

Heart Weight

Effect plot for D6MIT149
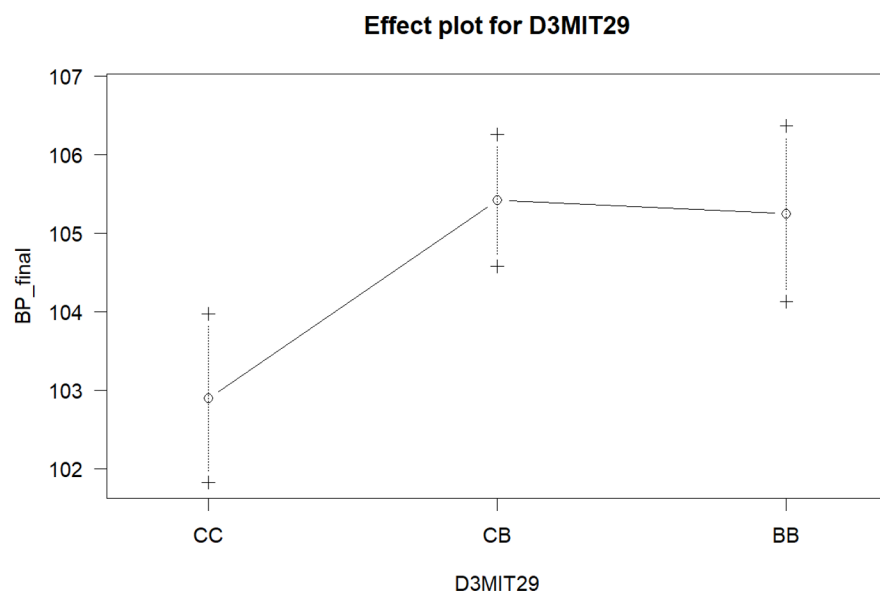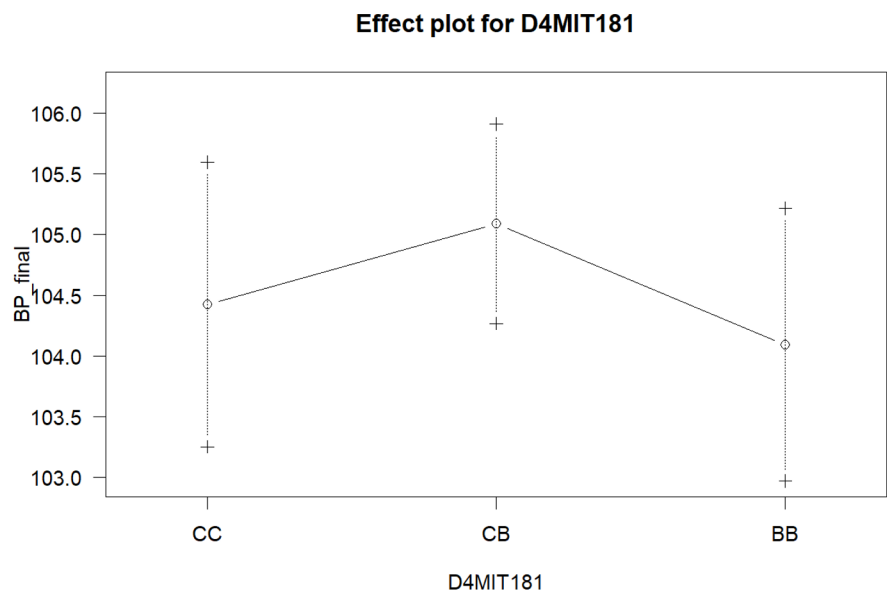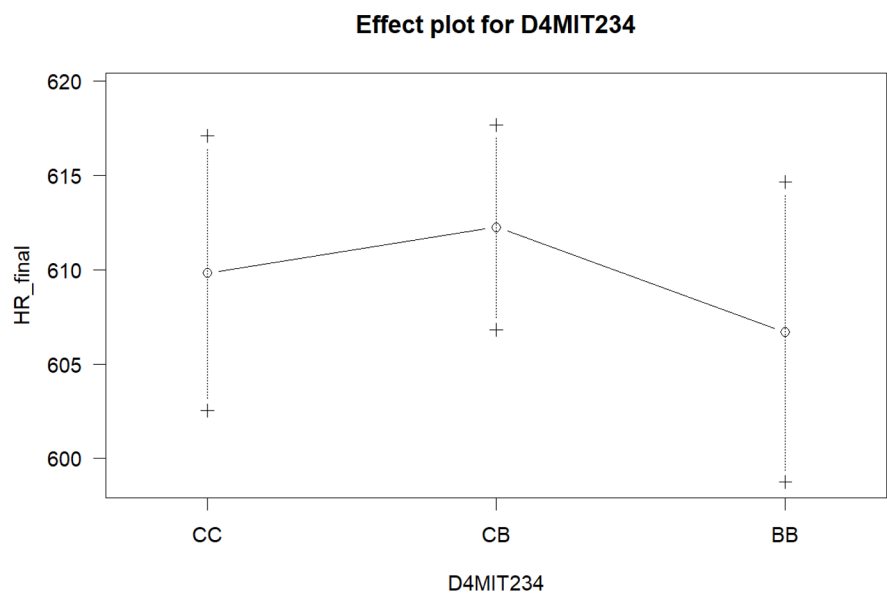
Narrative Analysis

Blood Pressure:
The main scan for blood pressure shows that the gene for blood pressure is very likely to be located on the seventh chromosome. The effect plot shows that mice with a CC genotype have lower blood pressure, while mice with the CB or BB genotypes have higher blood pressure. We can thus assume that the C gene corresponds to lower blood pressure, and that the B gene corresponds to higher blood pressure.

Heart Rate:
The main scan for heart rate shows that the gene for heart rate is very likely to be located on the second chromosome. The effect plot shows that mice with the CC or CB genotypes have higher heart rates, while mice with the BB genotypes have lower heart rates. We can thus assume that the C gene corresponds to higher heart rates, and that the B gene corresponds to lower heart rates.
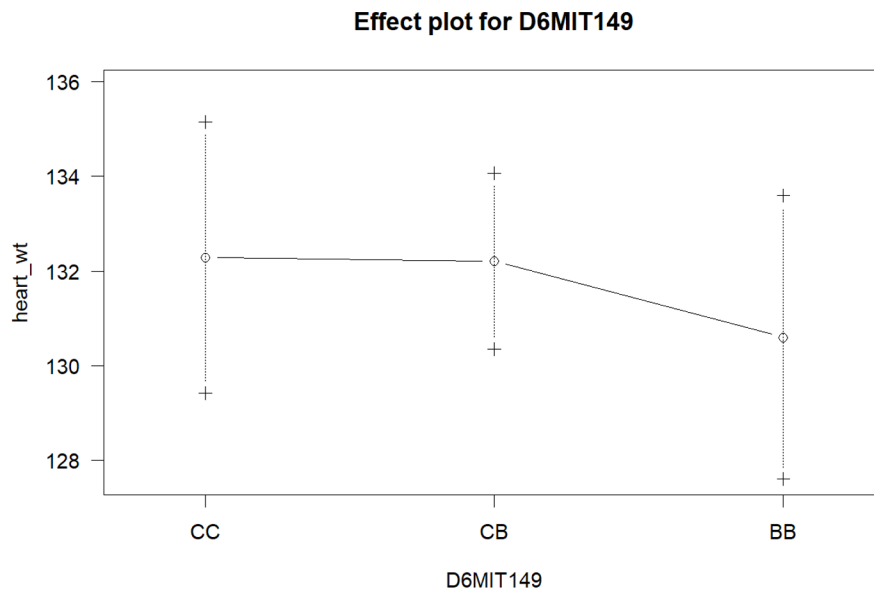
Heart Weight:
The main scan for heart rate shows that the gene for heart rate is very likely to be located on the twelfth chromosome. The effect plot shows that mice with a CC or CB genotypes have higher heart weights, while mice with the BB genotype have lower heart weights. We can thus assume that the C gene corresponds to higher heart weights, and that the B gene corresponds to lower heart weights.

R/qtl Code

```
# R script for analyzing Sugiyama QTL data
# Arav Bhardwaj
# BhardwajSugiyama.R script
# October 15, 2022

# Clean things up
rm(list=ls())
setwd("/Users/abhardwaj24/Desktop/RFolder")

# Install and load QTL package
install.packages("qtl")
library(qtl)

# Load in data
cross <- read.cross("csv", file = "sugiyamashort.csv", genotypes = c("C", "H", "B"),
                    na.strings = "-",alleles = c("C", "B"))

# Get summary statistics
summary(cross)

# HISTOGRAMS #
# Blood pressure
bp <- cross$pheno$BP_final
hist(bp, xlab = "BP", main = "Blood Pressure")
# Heart rate
hr <- cross$pheno$HR_final
hist(hr, xlab = "Heart Rate", main = "Heart Rate")
# Heart Weight
hw <- cross$pheno$heart_wt
hist(hw, xlab = "Heart Weight", main = "Heart Weight")

# Genetic map and missing data
plot.map(cross)
plotMissing(cross)

# QQ PLOTS #
# Blood pressure
qqnorm(bp, main = "Blood Pressure")
qqline(bp)

# Heart rate
qqnorm(hr, main = "Heart Rate")
qqline(hr)

# Heart weight
qqnorm(hw, main = "Heart Weight")
qqline(hw)

# CROSS CALCS #
cross <- calc.genoprob(cross, step = 2.0, off.end = 0, error.prob = 1.0e-4,
                       map.function = "haldane", stepwidth = "fixed")

cross <- sim.geno(cross, step = 2.0, off.end = 0, error.prob = 1.0e-4,
                  map.function = "haldane", stepwidth = "fixed")
```

```
# MAIN SCANS #
# Blood pressure
cross.scanBP <- scanone(cross, pheno.col = 3, model = "normal", method = "em")
cross.scanBP.perm <- scanone(cross, pheno.col = 3, model = "normal", method = "em", n.perm = 100)
# Heart rate
cross.scanHR <- scanone(cross, pheno.col = 4, model = "normal", method = "em")
cross.scanHR.perm <- scanone(cross, pheno.col = 4, model = "normal", method = "em", n.perm = 100)
# Heart weight
cross.scanHW <- scanone(cross, pheno.col = 6, model = "normal", method = "em")
cross.scanHW.perm <- scanone(cross, pheno.col = 6, model = "normal", method = "em", n.perm = 100)

# PLOT MAIN SCANS #
# Blood pressure
plot(cross.scanBP, main="Blood Pressure")
threshBP <- summary(cross.scanBP.perm, alpha = c(0.37, 0.10, 0.05))
abline(h = threshBP[1], col = "blue")
abline(h = threshBP[2], col = "red")
abline(h = threshBP[3], col = "green")
# Heart rate
plot(cross.scanHR, main="Heart Rate")
threshHR <- summary(cross.scanHR.perm, alpha = c(0.37, 0.10, 0.05))
abline(h = threshHR[1], col = "blue")
abline(h = threshHR[2], col = "red")
abline(h = threshHR[3], col = "green")
# Heart weight
plot(cross.scanHW, main="Heart Weight")
threshHW <- summary(cross.scanHW.perm, alpha = c(0.37, 0.10, 0.05))
abline(h = threshHW[1], col = "blue")
abline(h = threshHW[2], col = "red")
abline(h = threshHW[3], col = "green")

# SUMMARY #
# Blood pressure
summary(cross.scanBP, perm = cross.scanBP.perm, alpha = 0.05)
# Heart rate
summary(cross.scanHR, perm = cross.scanHR.perm, alpha = 0.05)
# Heart weight
summary(cross.scanHW, perm = cross.scanHW.perm, alpha = 0.05)

# EFFECT PLOTS #
# Blood pressure
firstBP <- find.marker(cross, chr = 3, pos = 48.7)
effectplot(cross, pheno.col = 3, mname1 = firstBP)
secondBP <- find.marker(cross, chr = 4, pos = 12.0)
effectplot(cross, pheno.col = 3, mname1 = secondBP)
# Heart rate
firstHR <- find.marker(cross, chr = 4, pos = 59.8)
effectplot(cross, pheno.col = 4, mname1 = firstHR)
# Heart weight
firstHW <- find.marker(cross, chr = 6, pos = 52.2)
effectplot(cross, pheno.col = 6, mname1 = firstHW)

# EOF
```