Practical 5 Statistical Genetics

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Question 1

```
library(data.table)

data <- fread("YRI6.raw", header = FALSE, sep = " ")

num_individuals <- nrow(data)
num_snps <- ncol(data) - 6

percentage_missing <- mean(is.na(data)) * 100

cat("Number of individuals:", num_individuals, "\n")
```

```
## Number of individuals: 85
```

```
cat("Number of SNPs:", num_snps, "\n")
```

```
## Number of SNPs: 56574
```

```
cat("Percentage of missing data:", percentage_missing, "%\n")
```

```
## Percentage of missing data: 0 %
```

Question 2

```
genomic_data <- data[2:nrow(data), 7:ncol(data)]
genomic_data[, (1:ncol(genomic_data)) := lapply(.SD, as.numeric), .SDcols = 1:ncol(genomic_data)]
shared_mean <- matrix(c(NA), nrow = num_individuals, ncol = num_individuals)
shared_sd <- matrix(c(NA), nrow = num_individuals, ncol = num_individuals)
for (i in 1:num_individuals) {
  for (j in 1:num_individuals) {
    shared <- numeric(0)
    for (k in 1:nrow(genomic_data)){
    shared <- c(shared, 2 - abs(as.matrix(genomic_data[k,i, with=FALSE]) - as.matrix(genomic_data[k,j, 
    }
    mean <- mean(shared)
    sd <- sd(shared)
    shared_mean[i,j] <- mean
    shared_sd[i,j] <- sd
  }
}

print(shared_mean[1:5, 1:5])
```

```
##          [,1]     [,2]     [,3]     [,4]     [,5]
## [1,] 2.000000 1.250000 1.190476 1.154762 1.273810
## [2,] 1.250000 2.000000 1.297619 1.476190 1.190476
## [3,] 1.190476 1.297619 2.000000 1.178571 1.107143
## [4,] 1.154762 1.476190 1.178571 2.000000 1.190476
## [5,] 1.273810 1.190476 1.107143 1.190476 2.000000
```

```r
print(shared_sd[1:5, 1:5])
```

```
##           [,1]      [,2]      [,3]      [,4]      [,5]
## [1,] 0.0000000 0.6376727 0.6489322 0.6492637 0.6828570
## [2,] 0.6376727 0.0000000 0.6166322 0.6300926 0.6489322
## [3,] 0.6489322 0.6166322 0.0000000 0.7140705 0.6769502
## [4,] 0.6492637 0.6300926 0.7140705 0.0000000 0.6489322
## [5,] 0.6828570 0.6489322 0.6769502 0.6489322 0.0000000
```

Question 3

```r
p0 <- matrix(c(NA), nrow = num_individuals, ncol = num_individuals)
p2 <- matrix(c(NA), nrow = num_individuals, ncol = num_individuals)
m <- matrix(c(NA), nrow = num_individuals, ncol = num_individuals)
for (i in 1:num_individuals) {
  for (j in 1:num_individuals) {
    shared <- numeric(0)
    for (k in 1:nrow(genomic_data)){
      shared <- c(shared, 2 - abs(as.matrix(genomic_data[k,i, with=FALSE]) - as.matrix(genomic_data[k,j
    }
    p0[i,j] <- sum(shared == 0)/length(shared)
    p2[i,j] <- sum(shared == 2)/length(shared)
    m[i,j] = 1 - p0[i,j] + p2[i,j]
  }
}
print(p0[1:5, 1:5])
```

```
##           [,1]       [,2]       [,3]       [,4]      [,5]
## [1,] 0.0000000 0.10714286 0.13095238 0.14285714 0.1309524
## [2,] 0.1071429 0.00000000 0.08333333 0.07142857 0.1309524
## [3,] 0.1309524 0.08333333 0.00000000 0.17857143 0.1785714
## [4,] 0.1428571 0.07142857 0.17857143 0.00000000 0.1309524
## [5,] 0.1309524 0.13095238 0.17857143 0.13095238 0.0000000
```

```r
print(p2[1:5, 1:5])
```

```
##           [,1]      [,2]      [,3]      [,4]      [,5]
## [1,] 1.0000000 0.3571429 0.3214286 0.2976190 0.4047619
## [2,] 0.3571429 1.0000000 0.3809524 0.5476190 0.3214286
## [3,] 0.3214286 0.3809524 1.0000000 0.3571429 0.2857143
## [4,] 0.2976190 0.5476190 0.3571429 1.0000000 0.3214286
## [5,] 0.4047619 0.3214286 0.2857143 0.3214286 1.0000000
```

```r
print(m[1:5, 1:5])
```

```
##          [,1]     [,2]     [,3]     [,4]     [,5]
## [1,] 2.000000 1.250000 1.190476 1.154762 1.273810
## [2,] 1.250000 2.000000 1.297619 1.476190 1.190476
## [3,] 1.190476 1.297619 2.000000 1.178571 1.107143
## [4,] 1.154762 1.476190 1.178571 2.000000 1.190476
## [5,] 1.273810 1.190476 1.107143 1.190476 2.000000
```

```r
print(shared_mean[1:5, 1:5])
```

```
##          [,1]     [,2]     [,3]     [,4]     [,5]
## [1,] 2.000000 1.250000 1.190476 1.154762 1.273810
## [2,] 1.250000 2.000000 1.297619 1.476190 1.190476
## [3,] 1.190476 1.297619 2.000000 1.178571 1.107143
## [4,] 1.154762 1.476190 1.178571 2.000000 1.190476
## [5,] 1.273810 1.190476 1.107143 1.190476 2.000000
```

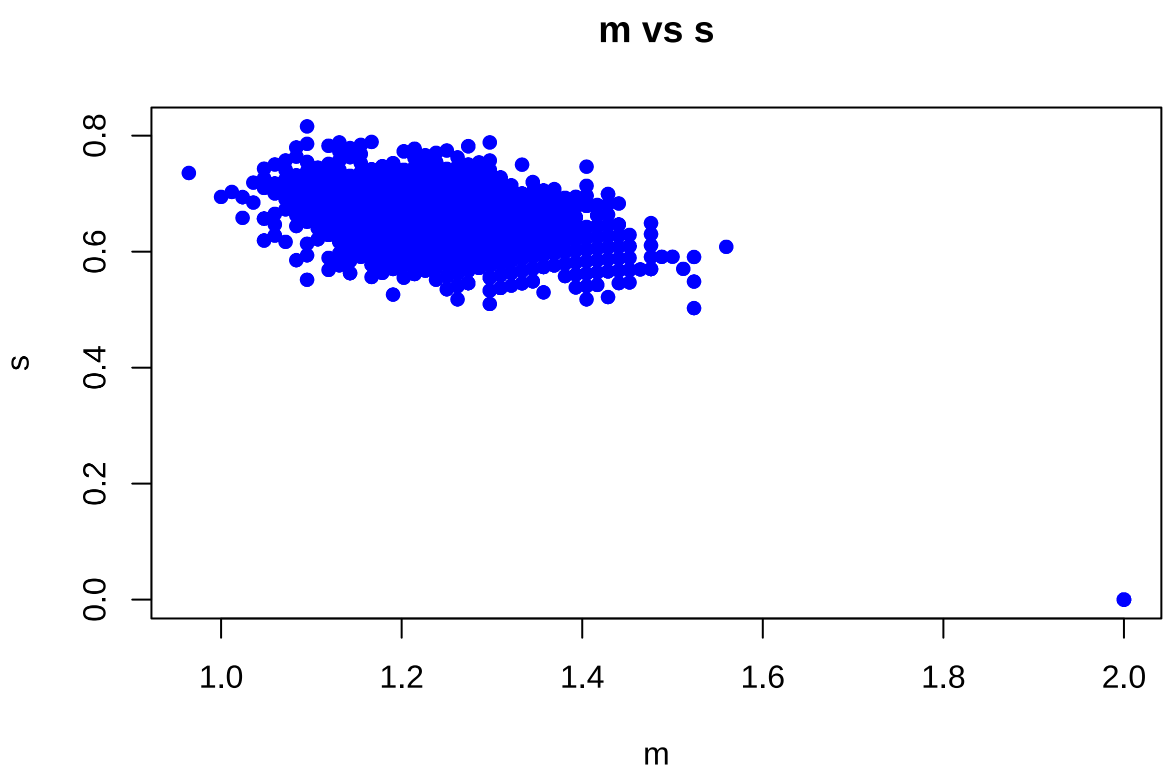
```
print(all.equal(shared_mean, m))
```

```
## [1] TRUE
```

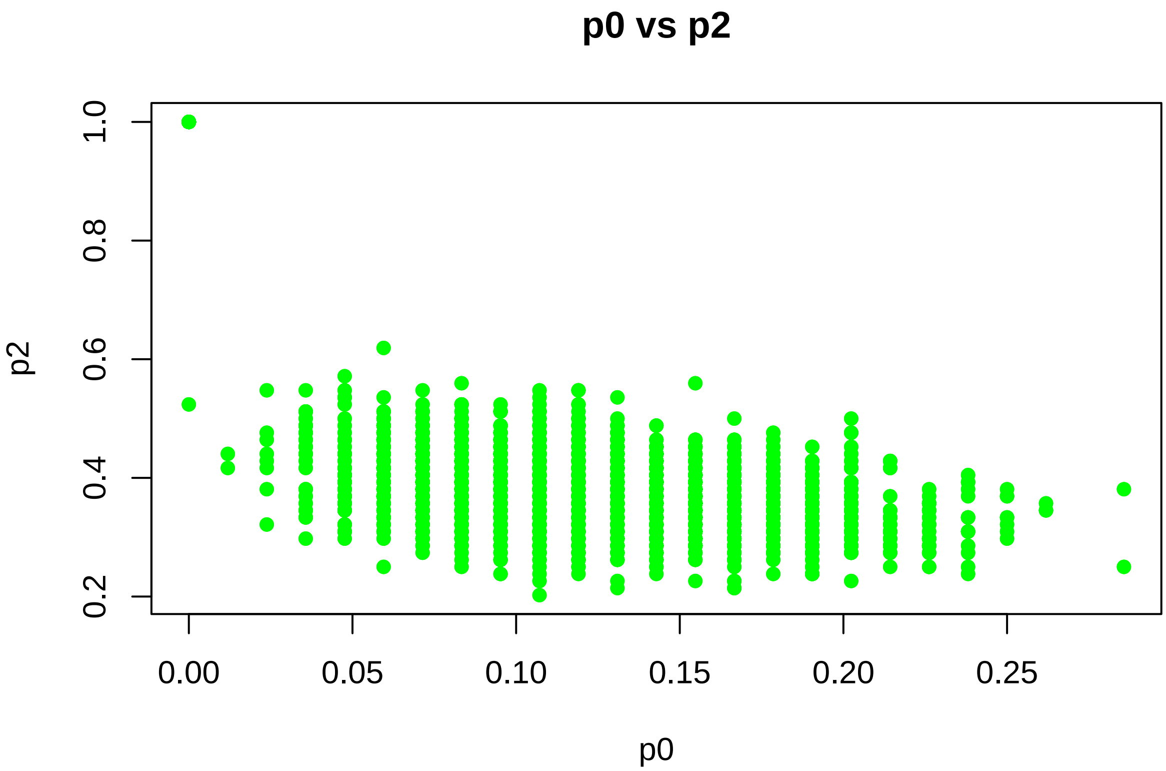
It holds because we can see that each element of m (m = 1 - p0 + p2) is equal to each element of shared_mean.

Question 4

```
plot(shared_mean, shared_sd, main = "m vs s", xlab = "m", ylab = "s", col = "blue", pch = 16)
```



```
plot(p0, p2, main = "p0 vs p2", xlab = "p0", ylab = "p2", col = "green", pch = 16)
```



m vs s : **Cluster in the Top Left:** The clustered points in this region could indicate a group of individuals with similar genetic characteristics. This grouping might be due to close familial relationships or specific genetic similarities.

Isolated Point in the Bottom Right: An isolated point suggests the presence of an individual or a small group of individuals with distinct genetic characteristics or differences from the majority. This could be due to unique genetic variations or atypical familial relationships.

p0 vs p2 : **Cluster on the Entire Bottom Part:** The concentration of points in the bottom part of the plot suggests that the majority of individuals share similar genetic characteristics in terms of *p0* and *p2*. This could result from genetic similarities within this population.

Isolated Point in the Top Left: The presence of an isolated point in the top left suggests that there is an individual or a group of individuals that stand out from the others in terms of *p0* and *p2*. This could indicate unique genetic variations or distinct features.

Question 5

```
m <- shared_mean
s <- shared_sd
family_relationship <- data[, c(3, 4)]
colors <- rep("blue", nrow(data) - 1)
parent_offspring_indices <- which(family_relationship[, 1] > 0 | family_relationship[, 2] > 0)
colors[parent_offspring_indices - 1] <- "red"
parent <- c()
for (i in 2:nrow(data)) {
```

```r
  print(data[i, 2] %in% family_relationship[[1]])
  if (data[i, 2] %in% family_relationship[[1]] | data[i, 2] %in% family_relationship[[2]]) {
    parent <- c(parent, i - 1)
  }
}
```

```
## [1] FALSE
## [1] FALSE
## [1] FALSE
## [1] TRUE
## [1] TRUE
## [1] FALSE
## [1] TRUE
## [1] FALSE
## [1] TRUE
## [1] FALSE
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## [1] FALSE
## [1] TRUE
## [1] TRUE
## [1] FALSE
## [1] TRUE
## [1] FALSE
## [1] FALSE
## [1] FALSE
```
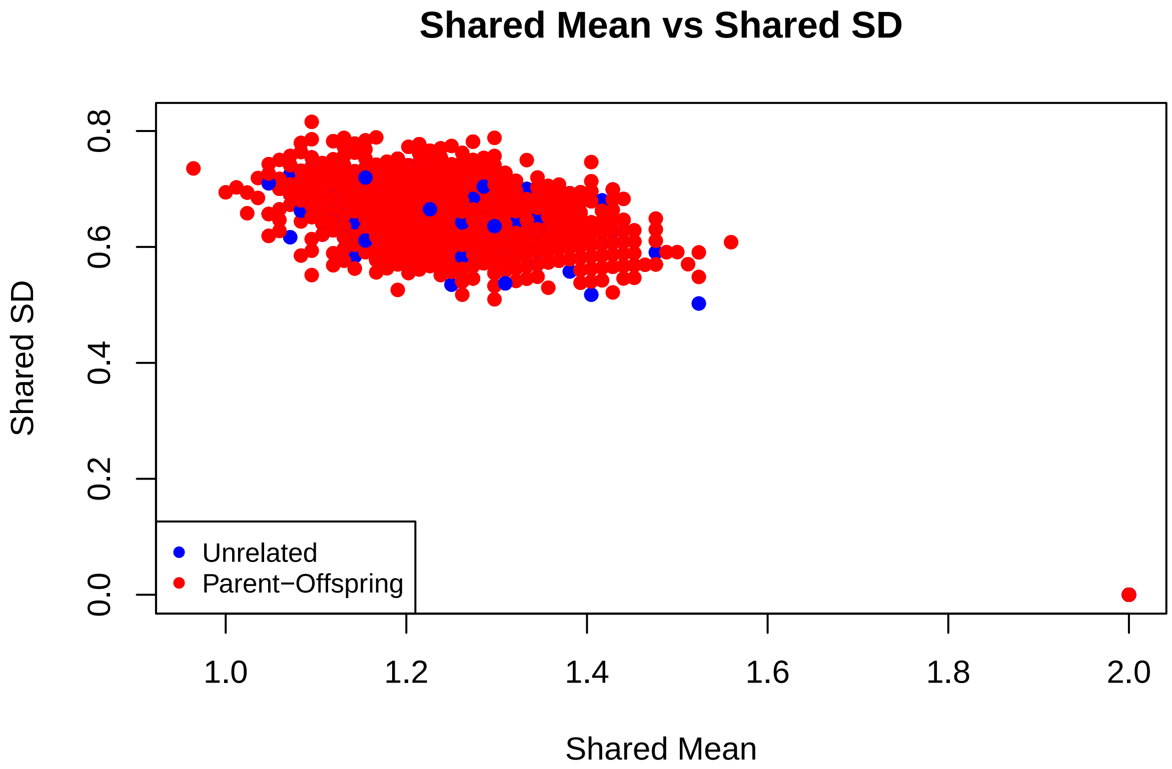

```
print(parent)
```

```
##  [1]  1  4  5  6  7  9 11 12 14 16 19 20 21 22 25 26 27 28 29 30 33 35 37 38 39
## [26] 43 44 46 48 49 51 52 53 55 57 58 59 61 64 65 66 67 69 71 74 76 77 78 79 81
## [51] 84
```

```
colors[parent] <- "red"

plot(m, s, main = "Shared Mean vs Shared SD", xlab = "Shared Mean", ylab = "Shared SD", col = colors, p
legend("bottomleft", legend = c("Unrelated", "Parent-Offspring"), col = c("blue", "red"), pch = 16, cex
```



We can see that all the majority of points present in the clusters have a family relationship. It affirms the hypothesis said before in question 4.