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Assigment BSG Stats 1

Fisrt data set

Question 1

```
file <-"TSICHR22RAW.raw"
data <- read.table(file, header=FALSE, sep=" ")
genetic_data <- data[, 7:ncol(data)]
first_elements <- sapply(genetic_data, function(x) x[1])
rs_columns <- grepl("^rs", first_elements)
genetic_data_rs <- genetic_data[, rs_columns]
print(dim(genetic_data_rs))
```

```
## [1]   103 20649
```

```
num_variants <- ncol(genetic_data_rs)
missing_percentage <- mean(is.na(genetic_data_rs)) * 100

cat("Number of variants in the database:", num_variants, "\n")
```

```
## Number of variants in the database: 20649
```

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

```
## Percentage of missing data: 0.1967231 %
```

Question 2

```
monomorphic_variants <- sapply(genetic_data_rs, function(col) {
  cleaned_col <- col[-1]  # Remove the first element (variant name)
  length(unique(cleaned_col[!is.na(cleaned_col)])) == 1
})
percentage_monomorphic <- (sum(monomorphic_variants) / length(monomorphic_variants)) * 100
non_monomorphic_data <- genetic_data_rs[, !monomorphic_variants]
num_remaining_variants <- ncol(non_monomorphic_data)

cat("Percentage of monomorphic variants:", percentage_monomorphic, "%\n")
```

```
## Percentage of monomorphic variants: 11.45818 %
```

```
cat("Number of remaining variants:", num_remaining_variants, "\n")
```

```
## Number of remaining variants: 18283
```

Question 3

```
column_index <- which(sapply(genetic_data_rs, function(col) col[1] == "rs8138488_C"))
rs8138488_column <- genetic_data_rs[, column_index]
genotype_counts <- table(rs8138488_column[-1])
genotype_0 <- genotype_counts["0"]
genotype_1 <- genotype_counts["1"]
```

```r
genotype_2 <- genotype_counts["2"]
minor_allele_count <- min(genotype_0, genotype_1, genotype_2)
total_individuals <- length(rs8138488_column) - 1
MAF <- minor_allele_count / (2 * total_individuals)
cat("Genotype counts for rs8138488_C:\n")
```

```
## Genotype counts for rs8138488_C:
```

```r
print(genotype_counts)
```

```
##
##  0  1  2
## 41 47 14
```

```r
cat("Minor Allele Count:", minor_allele_count, "\n")
```

```
## Minor Allele Count: 14
```

```r
cat("Minor Allele Frequency (MAF):", MAF, "\n")
```

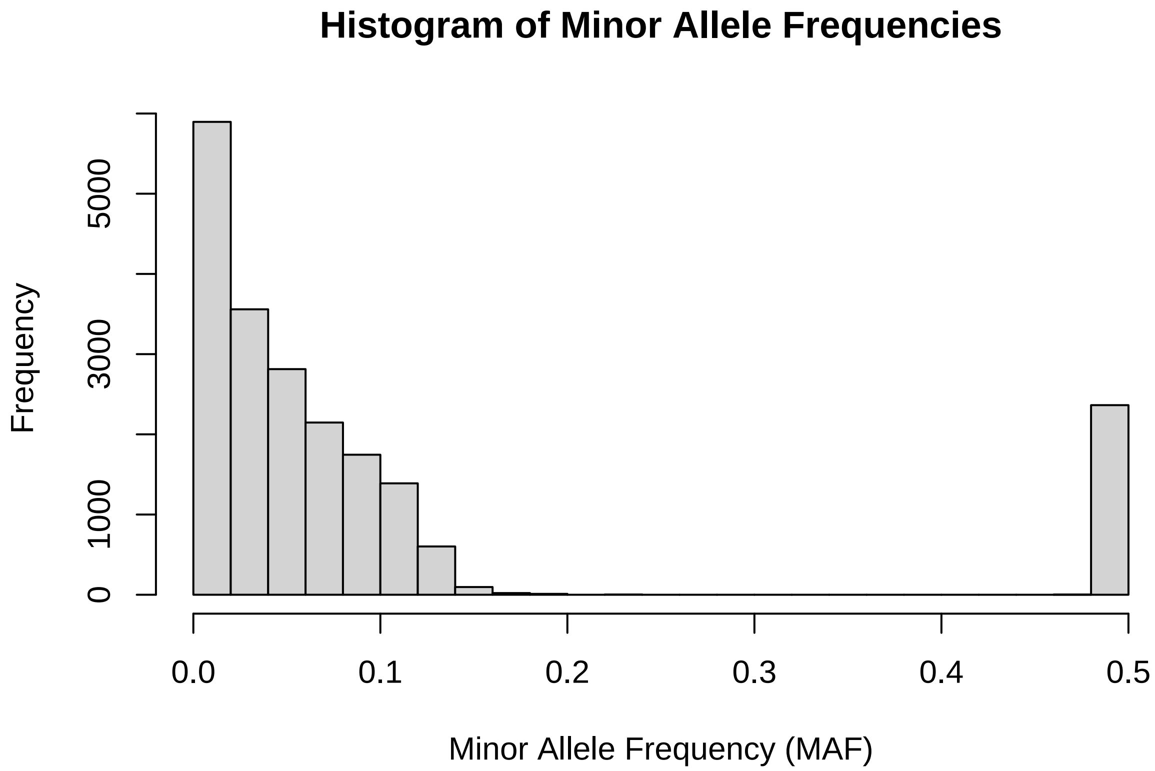
```
## Minor Allele Frequency (MAF): 0.06862745
```

Question 4

```r
MAF_values <- numeric(length(genetic_data_rs))

for (i in 1:length(genetic_data_rs)) {
  column <- genetic_data_rs[[i]]
  allele_counts <- table(column[-1])
  minor_allele_count <- min(allele_counts)
  total_individuals <- length(column) - 1
  MAF <- minor_allele_count / (2 * total_individuals)
  MAF_values[i] <- MAF
}

hist(MAF_values, breaks = 20, xlab = "Minor Allele Frequency (MAF)", main = "Histogram of Minor Allele
```



```
percentage_below_005 <- sum(MAF_values < 0.05) / length(MAF_values) * 100
percentage_below_001 <- sum(MAF_values < 0.01) / length(MAF_values) * 100

cat("Percentage of markers with MAF below 0.05:", percentage_below_005, "%\n")
```

```
## Percentage of markers with MAF below 0.05: 52.87423 %
```

```
cat("Percentage of markers with MAF below 0.01:", percentage_below_001, "%\n")
```

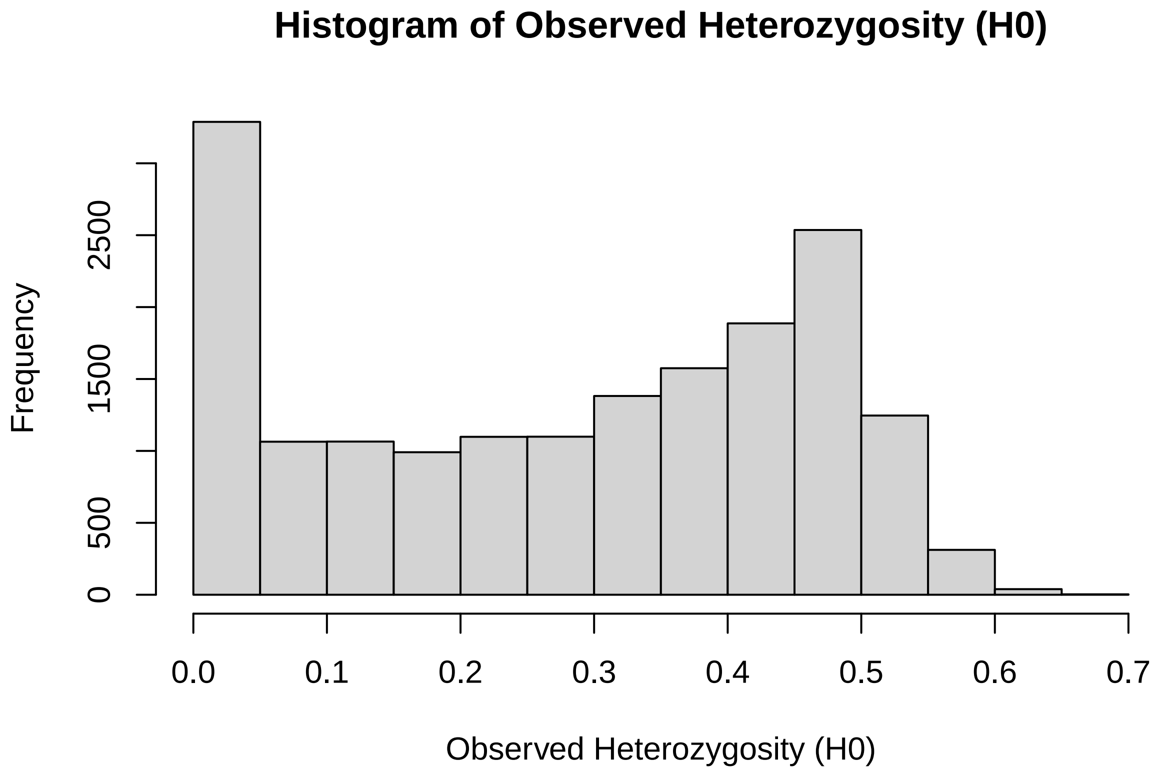
```
## Percentage of markers with MAF below 0.01: 17.74904 %
```

Question 5

```
HO_values <- numeric(length(genetic_data_rs))

for (i in 1:length(genetic_data_rs)) {
  column <- genetic_data_rs[[i]]
  genotypes <- column[-1]
  num_heterozygous <- sum(genotypes == 1)
  total_individuals <- length(genotypes)
  HO <- num_heterozygous / total_individuals
  HO_values[i] <- HO
}

hist(HO_values, breaks = 20, xlab = "Observed Heterozygosity (HO)", main = "Histogram of Observed Heter
```



Theoretical range of variation for H0 is [0, 0.5]

Question 6

```
library(HardyWeinberg)
```

```
## Loading required package: mice

##
## Attaching package: 'mice'

## The following object is masked from 'package:stats':
##
##     filter

## The following objects are masked from 'package:base':
##
##     cbind, rbind

## Loading required package: Rsolnp

## Loading required package: nnet
```

```
data(NistSTRs)
dimensions <- dim(NistSTRs)
num_individuals <- dimensions[1]
num_STRs <- dimensions[2]
cat("Number of individuals in the database:", num_individuals, "\n")
```

```
## Number of individuals in the database: 361
```

```r
cat("Number of STRs in the database:", num_STRs, "\n")
```

```
## Number of STRs in the database: 58
```

Second data set

Question 1

```r
library(HardyWeinberg)
data(NistSTRs)
dimensions <- dim(NistSTRs)
num_individuals <- dimensions[1]
num_STRs <- dimensions[2]
cat("Number of individuals in the database:", num_individuals, "\n")
```

```
## Number of individuals in the database: 361
```

```r
cat("Number of STRs in the database:", num_STRs, "\n")
```

```
## Number of STRs in the database: 58
```

Question 2

```r
count_alleles <- function(STR_locus) {
  unique_alleles <- unique(STR_locus)
  num_alleles <- length(unique_alleles)
  return(num_alleles)
}
num_alleles_list <- sapply(NistSTRs, count_alleles)

mean_num_alleles <- mean(num_alleles_list)
sd_num_alleles <- sd(num_alleles_list)
median_num_alleles <- median(num_alleles_list)
min_num_alleles <- min(num_alleles_list)
max_num_alleles <- max(num_alleles_list)

cat("Descriptive statistics of the number of alleles:\n")
```

```
## Descriptive statistics of the number of alleles:
```

```r
cat("Mean:", mean_num_alleles, "\n")
```

```
## Mean: 9.482759
```

```r
cat("Standard Deviation:", sd_num_alleles, "\n")
```

```
## Standard Deviation: 5.006106
```

```r
cat("Median:", median_num_alleles, "\n")
```

```
## Median: 8
```

```r
cat("Minimum:", min_num_alleles, "\n")
```

```
## Minimum: 5
```

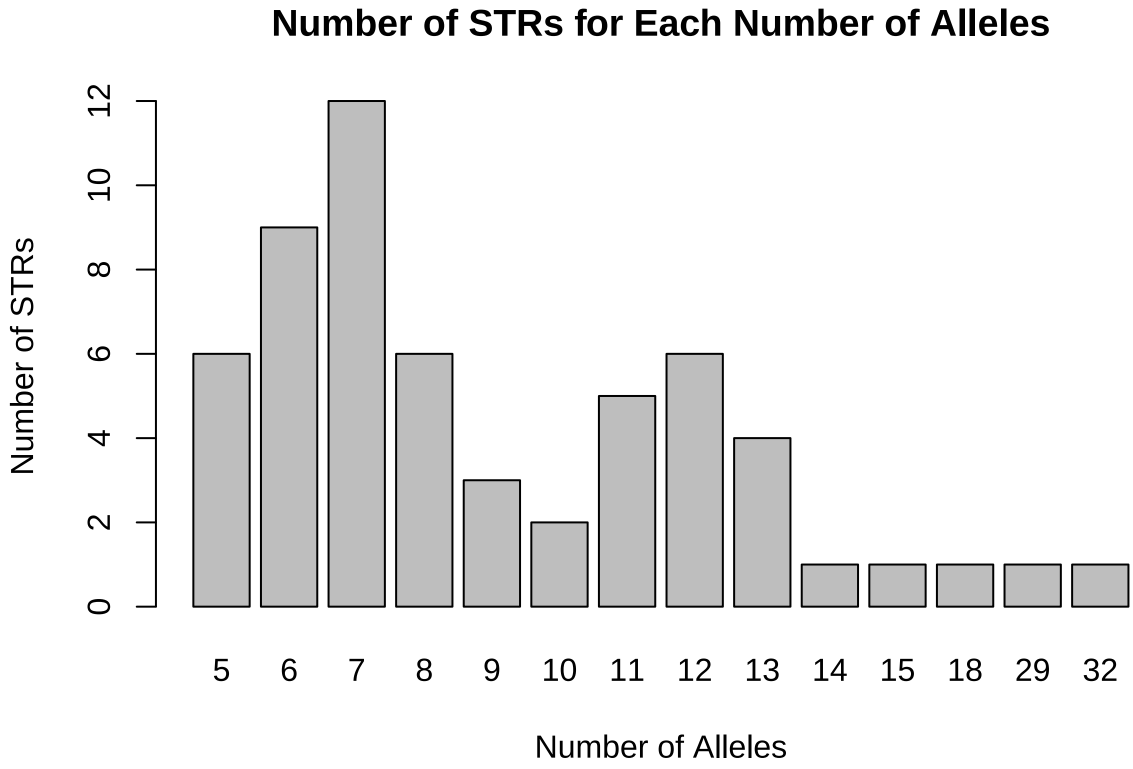
```
cat("Maximum:", max_num_alleles, "\n")
```

```
## Maximum: 32
```

Question 3

```
table_num_alleles <- table(num_alleles_list)

barplot(table_num_alleles, xlab = "Number of Alleles", ylab = "Number of STRs", main = "Number of STRs f
```



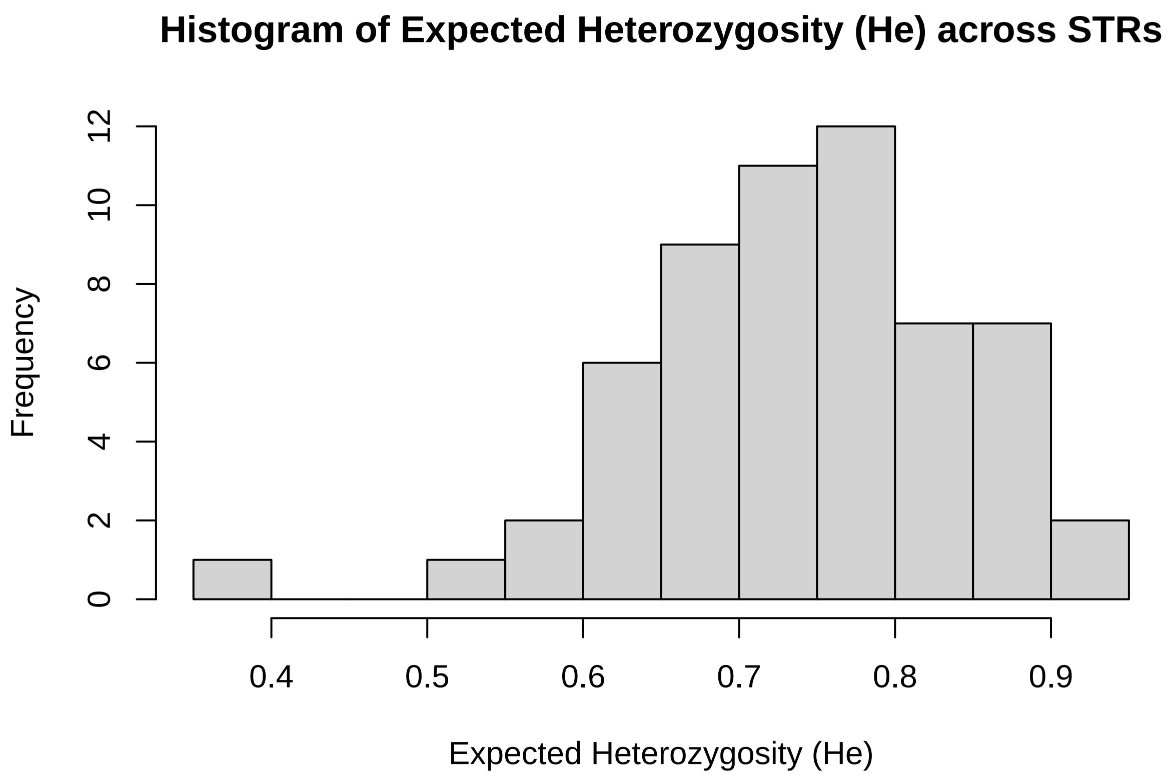
```
most_common_alleles <- names(table_num_alleles)[which.max(table_num_alleles)]
cat("The most common number of alleles for an STR is:", most_common_alleles, "\n")
```

```
## The most common number of alleles for an STR is: 7
```

Question 4

```
calculate_He <- function(STR_locus) {
  unique_alleles <- unique(STR_locus)
  allele_freq <- table(STR_locus) / length(STR_locus)
  He <- 1 - sum((allele_freq / sum(allele_freq))^2)
  return(He)
}
He_values <- sapply(NistSTRs, calculate_He)

hist(He_values, breaks = 20, xlab = "Expected Heterozygosity (He)", main = "Histogram of Expected Hetero
```



```
average_He <- mean(He_values)
cat("Average Expected Heterozygosity over all STRs:", average_He, "\n")
```

```
## Average Expected Heterozygosity over all STRs: 0.7404483
```

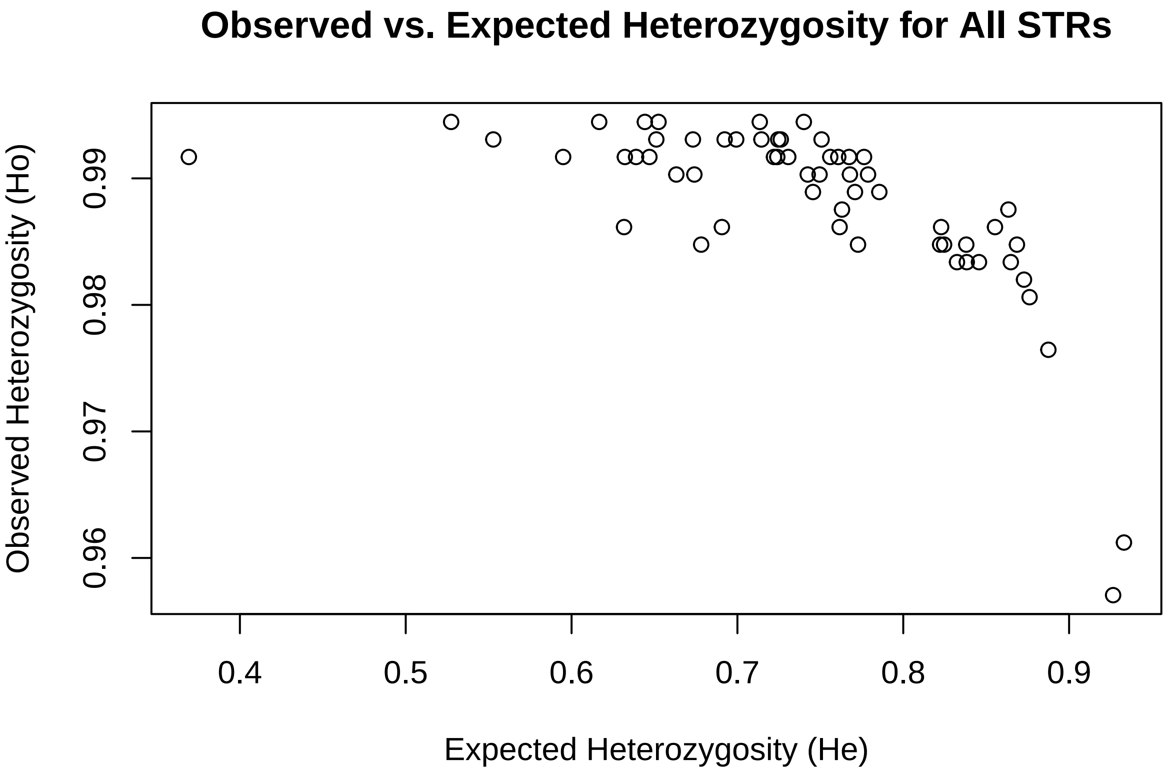
Question 5

```
calculate_Ho <- function(STR_locus) {
  num_unique_alleles <- length(unique(STR_locus))
  total_individuals <- length(STR_locus)
  Ho <- 1 - (num_unique_alleles - 1) / (2 * total_individuals)
  return(Ho)
}
Ho_values <- sapply(NistSTRs, calculate_Ho)

heterozygosity_data <- data.frame(He = He_values, Ho = Ho_values)
print(heterozygosity_data)
```

```
##                    He        Ho
## CSF1PO-1    0.6731686 0.9930748
## CSF1PO-2    0.6441786 0.9944598
## D10S1248-1 0.6511000 0.9930748
## D10S1248-2 0.7608137 0.9916898
## D12S391-1  0.8456657 0.9833795
## D12S391-2  0.8761750 0.9806094
## D13S317-1  0.7507462 0.9930748
## D13S317-2  0.7424130 0.9903047
## D16S539-1  0.7245954 0.9930748
## D16S539-2  0.6992733 0.9930748
## D18S51-1   0.8324061 0.9833795
## D18S51-2   0.8553188 0.9861496
## D19S433-1  0.6906331 0.9861496
## D19S433-2  0.7727227 0.9847645
## D1S1656-1  0.8648491 0.9833795
## D1S1656-2  0.8728141 0.9819945
## D21S11-1   0.7616271 0.9861496
## D21S11-2   0.8246253 0.9847645
## D22S1045-1 0.7220479 0.9916898
## D22S1045-2 0.5949156 0.9916898
## D2S1338-1  0.8228298 0.9861496
## D2S1338-2  0.8634065 0.9875346
## D2S441-1   0.6631625 0.9903047
## D2S441-2   0.7455437 0.9889197
## D3S1358-1  0.7240583 0.9916898
## D3S1358-2  0.7559948 0.9916898
## D5S818-1   0.6388993 0.9916898
## D5S818-2   0.6321314 0.9916898
## D6S1043-1  0.6781409 0.9847645
## D6S1043-2  0.8685477 0.9847645
## D7S820-1   0.7763599 0.9916898
## D7S820-2   0.7673360 0.9916898
## D8S1179-1  0.7855679 0.9889197
## D8S1179-2  0.7678425 0.9903047
## F13A01-1   0.7134844 0.9944598
## F13A01-2   0.6316403 0.9861496
## F13B-1     0.7400496 0.9944598
## F13B-2     0.5528042 0.9930748
## FESFPS-1   0.6166466 0.9944598
## FESFPS-2   0.6524351 0.9944598
## FGA-1      0.8222159 0.9847645
## FGA-2      0.8379923 0.9847645
## LPL-1      0.5273747 0.9944598
## LPL-2      0.6922906 0.9930748
## Penta_C-1  0.6740433 0.9903047
## Penta_C-2  0.7306574 0.9916898
## Penta_D-1  0.7630543 0.9875346
## Penta_D-2  0.7708965 0.9889197
## Penta_E-1  0.8382993 0.9833795
## Penta_E-2  0.8875009 0.9764543
## SE33-1     0.9330806 0.9612188
## SE33-2     0.9265429 0.9570637
## TH01-1     0.7261608 0.9930748
## TH01-2     0.6469410 0.9916898
## TPOX-1     0.3692114 0.9916898
## TPOX-2     0.7144666 0.9930748
## vWA-1      0.7787693 0.9903047
## vWA-2      0.7495338 0.9903047
```

```
plot(heterozygosity_data$He, heterozygosity_data$Ho, xlab = "Expected Heterozygosity (He)", ylab = "Obse
     main = "Observed vs. Expected Heterozygosity for All STRs")
```


Observed vs. Expected Heterozygosity for All STRs
Observed Heterozygosity (Ho)
0.96
0.97
0.98
0.99
0.4
0.5
0.6
0.7
0.8
0.9
Expected Heterozygosity (He)