

answers.r

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```
# Question 1
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

library(data.table)

file <- "TSIChr22v4.raw"
data <- fread(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, with = FALSE]
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: 1101343

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

## Percentage of missing data: 0 %

# 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, with = FALSE]
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num_remaining_variants <- ncol(non_monomorphic_data)
cat("Percentage of monomorphic variants:", percentage_monomorphic, "%\n")

## Percentage of monomorphic variants: 81.0347 %
cat("Number of remaining variants:", num_remaining_variants, "\n")

## Number of remaining variants: 208873
# Question 3

variant_column_index <- which(genetic_data_rs[1, ] == "rs587756191_T")
variant_column <- genetic_data_rs[[variant_column_index]]

variant_column <- factor(variant_column, levels = c(0, 1, 2), labels = c("AA", "AB", "BB"))
print(table(variant_column))

## variant_column
## AA AB BB
## 106 1 0
# Create a table with counts
chi_square_result <- HWChisq(table(variant_column))

## Warning in HWChisq(table(variant_column)): Expected counts below 5: chi-square
## approximation may be incorrect

## Chi-square test with continuity correction for Hardy-Weinberg equilibrium (autosomal)
## Chi2 = 106.2512 DF = 1 p-value = 6.495738e-25 D = 0.002336449 f = -0.004694836
print(chi_square_result)

## $chisq
## [1] 106.2512
##
## $pval
## [1] 6.495738e-25
##
## $D
## [1] 0.002336449
##
## $p
## [1] 0.004672897
##
## $f
## [1] -0.004694836
##
## $expected
## AA AB BB
## 2.336449e-03 9.953271e-01 1.060023e+02
##
## $chi.contrib
## AA AB BB
## 1.060023e+02 2.465008e-01 2.336449e-03
# Chi-square test with continuity correction
chi_square_correction_result <- HWChisq(table(variant_column))

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## Warning in HWChisq(table(variant_column)): Expected counts below 5: chi-square
## approximation may be incorrect

## Chi-square test with continuity correction for Hardy-Weinberg equilibrium (autosomal)
## Chi2 = 106.2512 DF = 1 p-value = 6.495738e-25 D = 0.002336449 f = -0.004694836
print(chi_square_correction_result)

## $chisq
## [1] 106.2512
##
## $pval
## [1] 6.495738e-25
##
## $D
## [1] 0.002336449
##
## $p
## [1] 0.004672897
##
## $f
## [1] -0.004694836
##
## $expected
##           AA           AB           BB
## 2.336449e-03 9.953271e-01 1.060023e+02
##
## $chi.contrib
##           AA           AB           BB
## 1.060023e+02 2.465008e-01 2.336449e-03

# Exact test
exact_test_result <- HWExact(table(variant_column))

## Haldane Exact test for Hardy-Weinberg equilibrium (autosomal)
## using SELOME p-value
## sample counts: nAA = 106 nAB = 1 nBB = 0
## H0: HWE (D=0), H1: D <> 0
## D = 0.002336449 p-value = 1
print(exact_test_result)

## $pval
## [1] 1
##
## $prob
## 1
## 1
##
## $pofthesample
## 1
## 1

# Permutation test
permutation_test_result <- HWPerm(table(variant_column))

## Permutation test for Hardy-Weinberg equilibrium
## Observed statistic: 0.002358439 17000 permutations. p-value: 1

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print(permutation_test_result)

## $stat
## [1] 0.002358439
##
## $pval
## [1] 1

# Question 4
genetic_data_rs <- non_monomorphic_data
genotype_counts_matrix <- matrix(NA, ncol = 3, nrow = ncol(genetic_data_rs))
for (i in 1:ncol(genetic_data_rs)) {
  variant_column <- genetic_data_rs[[i]]
  genotype_counts <- table(variant_column)

  genotype_counts <- genotype_counts[c("0", "1", "2")]

  genotype_counts_matrix[i, ] <- as.numeric(genotype_counts)
}

colnames(genotype_counts_matrix) <- c("AA", "AB", "BB")

# Question 5
genetic_data_no_header <- genetic_data_rs[-1, ]

hwe_results <- c()

# Loop through each SNP column
for (col_name in names(genetic_data_no_header)) {
  # Extract the SNP column
  variant_column <- as.integer(genetic_data_no_header[[col_name]])
  variant_column <- factor(variant_column, levels = c(0, 1, 2), labels = c("AA", "AB", "BB"))

  # Create a matrix X with genotype counts for the SNP
  X <- matrix(table(variant_column), ncol = 3, byrow = TRUE)

  # Apply HWExactStats to the matrix X
  hwe_result <- HWExactStats(X, x.linked = FALSE, plinkcode = TRUE, midp = FALSE)

  # Collect the p-value for each SNP
  hwe_results <- c(hwe_results, hwe_result)
}

# Calculate the percentage of significant SNPs (p-value < 0.05)
significant_snps_percentage <- mean(hwe_results < 0.05) * 100

cat("Percentage of significant SNPs at alpha = 0.05:", significant_snps_percentage, "%\n")

## Percentage of significant SNPs at alpha = 0.05: 2.761965 %

# Question 6

most_significant_index <- which.min(hwe_results)

most_significant_snp_value <- genetic_data_rs[[most_significant_index]][1]

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most_significant_p_value <- hwe_results[most_significant_index]

cat("Most Significant SNP:", most_significant_snp_value, "\n")

## Most Significant SNP: rs2629366_C
cat("P-value for Most Significant SNP:", most_significant_p_value, "\n")

## P-value for Most Significant SNP: 9.784766e-33
# Question 7

inbreeding_coefficients <- c()

# Loop through each SNP column
for (col_name in names(genetic_data_no_header)) {
  # Extract the SNP column
  variant_column <- as.integer(genetic_data_no_header[[col_name]])
  variant_column <- factor(variant_column, levels = c(0, 1, 2), labels = c("AA", "AB", "BB"))

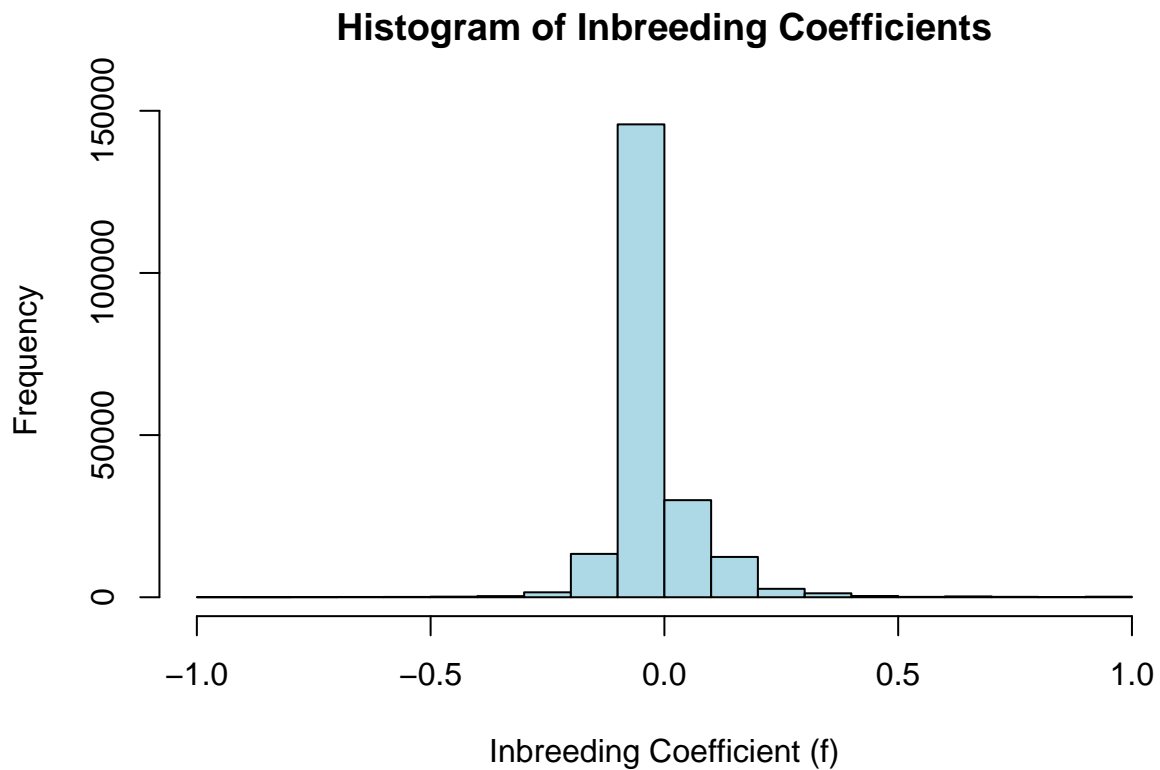
  # Create a matrix X with genotype counts for the SNP
  X <- matrix(table(variant_column), ncol = 3, byrow = TRUE)

  # Apply HWf to calculate the inbreeding coefficient
  f <- HWf(X)

  # Store the inbreeding coefficient
  inbreeding_coefficients <- c(inbreeding_coefficients, f)
}

# Make a histogram of f
hist(inbreeding_coefficients, main = "Histogram of Inbreeding Coefficients", xlab = "Inbreeding Coefficient")

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# Calculate and print descriptive statistics
mean_f <- mean(inbreeding_coefficients)
sd_f <- sd(inbreeding_coefficients)

cat("Descriptive Statistics for Inbreeding Coefficients:\n")

## Descriptive Statistics for Inbreeding Coefficients:
cat("Mean:", mean_f, "\n")

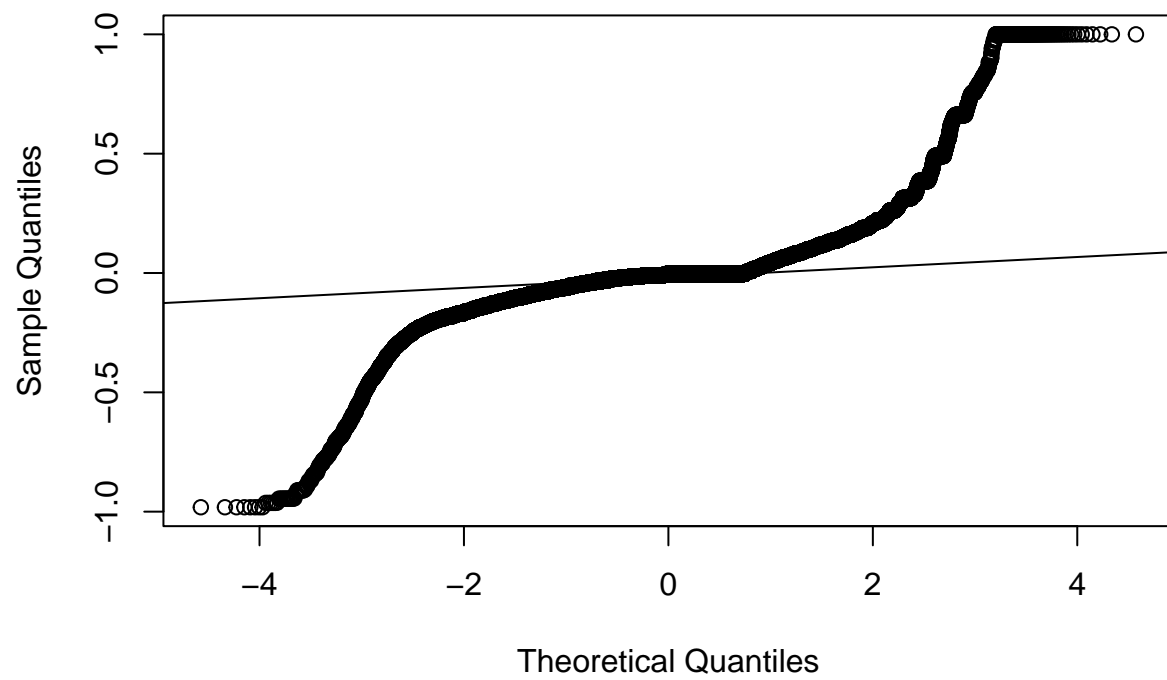
## Mean: -0.004668641

cat("Standard Deviation:", sd_f, "\n")

## Standard Deviation: 0.09486517

# Probability plot
qqnorm(inbreeding_coefficients, main = "Probability Plot of Inbreeding Coefficients")
qqline(inbreeding_coefficients)
```

Probability Plot of Inbreeding Coefficients



Question 8