

# 509HW2

## Quarto

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## Running Code

When you click the **Render** button a document will be generated that includes both content and the output of embedded code. You can embed code like this:

```
1 + 1
```

```
[1] 2
```

You can add options to executable code like this

```
[1] 4
```

The `echo: false` option disables the printing of code (only output is displayed).

## 1. Numbers of genes on each chromosome

(20%) Visualize using bar plot (function `barplot()`) the total number of genes on each chromosome (1-22, X, Y). Report the chromosomes with the min and max number of genes.

```
# import GFF3 table
library(stringr)
file <- "E:/Language/R/gencode.v44.primary_assembly.annotation.gff3"
GFF <- read.table(file, header = FALSE, sep = "\t")
```

```

colnames(GFF) <- c("seqid", "source", "type", "start", "end",
                  "score", "strand", "phase", "attributes")
rownames(GFF) <- 1:nrow(GFF)

# Visualize and calculate minimum and maximum values

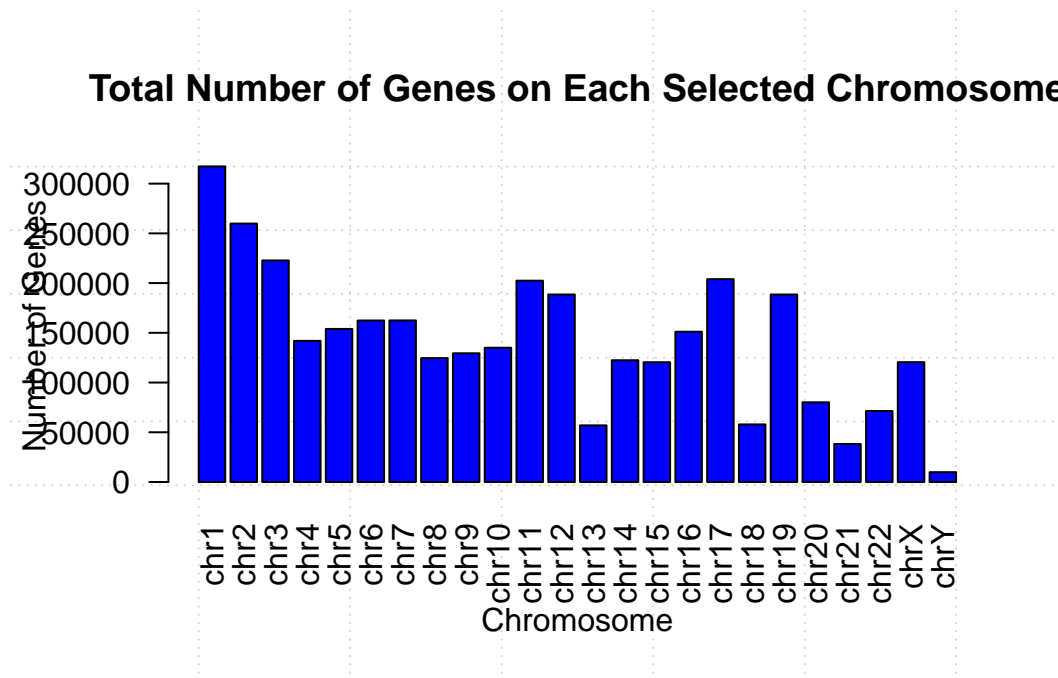
chromosome_counts <- table(GFF$seqid)

# Select only data for chr1 to chr22, X, and Y chromosomes
selected_chromosomes <- c("chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8",
                          "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "
                          "chr20", "chr21", "chr22", "chrX", "chrY")

chromosome_counts <- chromosome_counts[selected_chromosomes]

# Create a larger bar plot to visualize gene counts
options(repr.plot.width = 10, repr.plot.height = 4) # Adjust the plot size as needed
barplot(chromosome_counts,
        names.arg = names(chromosome_counts),
        main = "Total Number of Genes on Each Selected Chromosome",
        xlab = "Chromosome",
        ylab = "Number of Genes",
        col = "blue",
        panel.first = grid(),
        las = 2)

```



```
# Calculate the chromosomes with the minimum and maximum number of genes
min_gene_chromosome <- names(chromosome_counts)[which.min(chromosome_counts)]
max_gene_chromosome <- names(chromosome_counts)[which.max(chromosome_counts)]

cat("Chromosome with the min number of genes:", min_gene_chromosome, "\n")
```

Chromosome with the min number of genes: chrY

```
cat("Chromosome with the max number of genes:", max_gene_chromosome, "\n")
```

Chromosome with the max number of genes: chr1

## 2. Gene density on each chromosome

(20%) Visualize the density of genes on each chromosome. Density is defined by the total number of genes divided by the length of the chromosome. You can use the maximum end coordinate value to approximate the length of a chromosome. Point out the chromosomes with the minimum and maximum gene density, respectively.

```

# Create a subset of GFF data for chr1-22, X, and Y chromosomes
selected_chromosomes <- c("chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8",
                          "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18",
                          "chr19", "chr20", "chr21", "chr22", "chrX", "chrY")

selected_GFF <- GFF[GFF$seqid %in% selected_chromosomes, ]

# Calculate gene density and Visualize and calculate min and max values
chromosome_lengths <- aggregate(selected_GFF$end, list(seqid = selected_GFF$seqid), max)
colnames(chromosome_lengths) <- c("seqid", "chromosome_length")

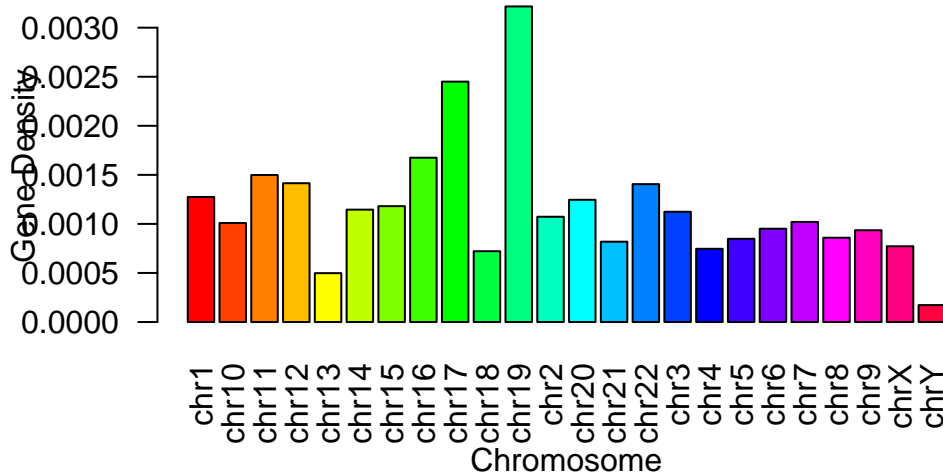
chromosome_gene_counts <- aggregate(selected_GFF$seqid, list(seqid = selected_GFF$seqid),
colnames(chromosome_gene_counts) <- c("seqid", "num_genes")

chromosome_gene_density <- merge(chromosome_lengths, chromosome_gene_counts, by = "seqid")
chromosome_gene_density$gene_density <- chromosome_gene_density$num_genes / chromosome_gene_density$chromosome_length

# Create a bar plot to visualize gene density
barplot(chromosome_gene_density$gene_density,
        names.arg = chromosome_gene_density$seqid,
        col = rainbow(nrow(chromosome_gene_density)), # Assign colors
        xlab = "Chromosome",
        ylab = "Gene Density",
        main = "Gene Density on Each Selected Chromosome",
        las = 2)

```

## Gene Density on Each Selected Chromosome



```
# Identify chromosomes with min and max gene density
min_density_chromosome <- chromosome_gene_density$seqid[which.min(chromosome_gene_density$
max_density_chromosome <- chromosome_gene_density$seqid[which.max(chromosome_gene_density$

cat("Chromosome with the min gene density:", min_density_chromosome, "\n")
```

Chromosome with the min gene density: chrY

```
cat("Chromosome with the max gene density:", max_density_chromosome, "\n")
```

Chromosome with the max gene density: chr19

### 3. Numbers and density of transcripts on each chromosome

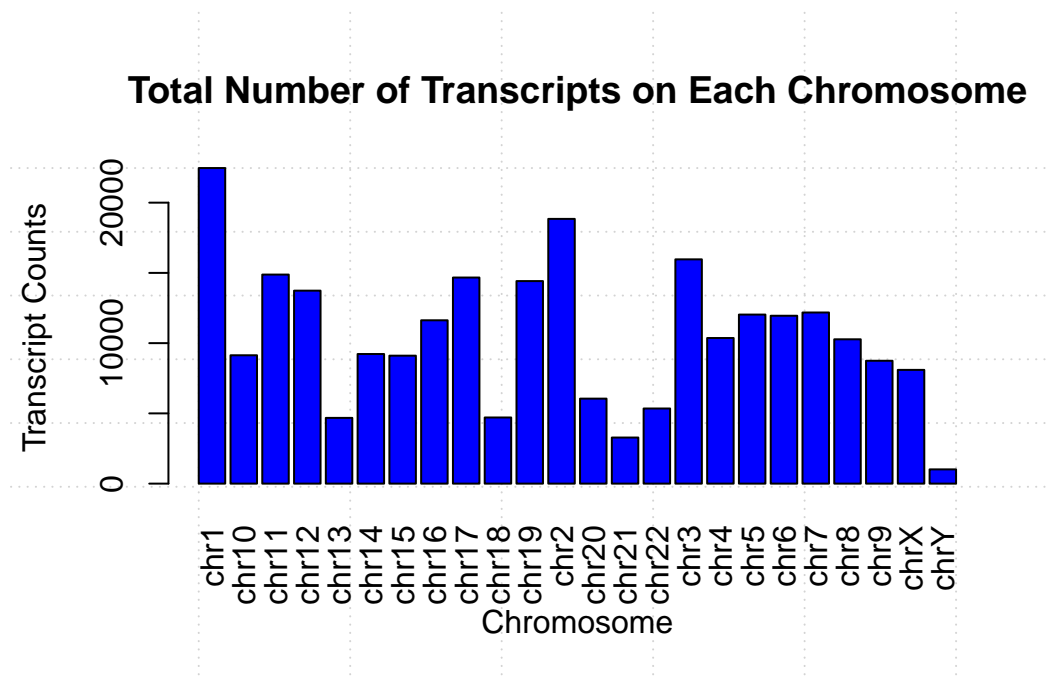
(20%) Repeat 1 and 2 for transcripts.

#### 3.1 Repeat 1

```
# Filter GFF data for selected chromosomes and transcript type
selected_GFF <- GFF[GFF$seqid %in% selected_chromosomes & GFF$type == "transcript", ]
```

```
# Calculate the number of transcripts for each chromosome
chromosome_transcript_counts <- table(selected_GFF$seqid)

# Create a larger bar plot to visualize transcript counts
options(repr.plot.width = 10, repr.plot.height = 4) # Adjust the plot size as needed
barplot(chromosome_transcript_counts,
        names.arg = names(chromosome_transcript_counts),
        main = "Total Number of Transcripts on Each Chromosome",
        xlab = "Chromosome",
        ylab = "Transcript Counts",
        col = "blue",
        panel.first=grid(),
        las = 3.5)
```



```
# Calculate the minimum and maximum
min_transcript_chromosome <- names(chromosome_transcript_counts)[which.min(chromosome_transcript_counts)]
max_transcript_chromosome <- names(chromosome_transcript_counts)[which.max(chromosome_transcript_counts)]

cat("Chromosome with the minimum transcript counts:", min_transcript_chromosome, "\n")
```

Chromosome with the minimum transcript counts: chrY

```
cat("Chromosome with the maximum transcript counts:", max_transcript_chromosome, "\n")
```

Chromosome with the maximum transcript counts: chr1

### 3.2 Repeat 2

```
# Calculate transcript density and visualize, and calculate min and max values
# Filter GFF to include only chr1-22 and X,Y chromosomes

chromosomes_of_interest <- c("chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8", "chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18", "chr19", "chr20", "chr21", "chr22", "X", "Y")
GFF_filtered <- GFF[GFF$seqid %in% chromosomes_of_interest, ]

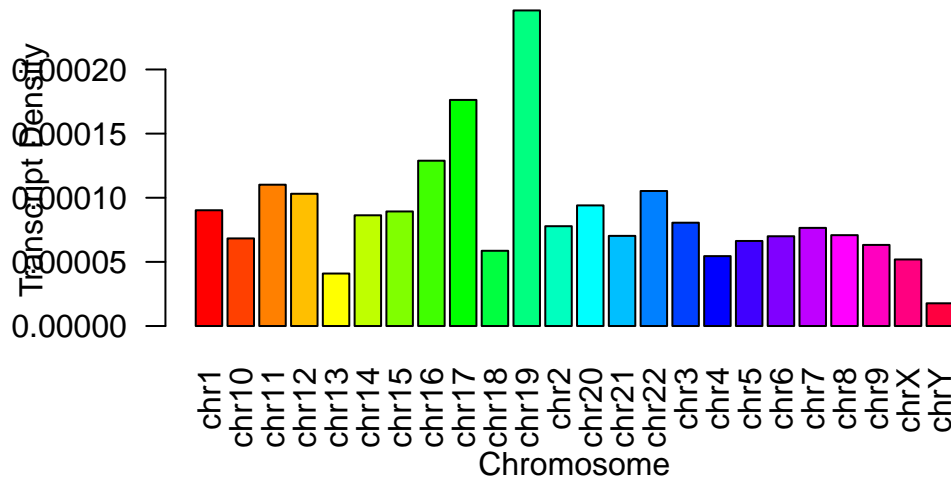
# Calculate transcript density and Visualize, and calculate min and max values
chromosome_lengths <- aggregate(GFF_filtered$end[GFF_filtered$type == "transcript"], list(chromosome_of_interest), FUN = sum)
colnames(chromosome_lengths) <- c("seqid", "chromosome_length")

chromosome_transcript_counts <- aggregate(GFF_filtered$seqid[GFF_filtered$type == "transcript"], list(chromosome_of_interest), FUN = sum)
colnames(chromosome_transcript_counts) <- c("seqid", "num_transcripts")

chromosome_transcript_density <- merge(chromosome_lengths, chromosome_transcript_counts, by = "seqid")
chromosome_transcript_density$transcript_density <- chromosome_transcript_density$num_transcripts / chromosome_transcript_density$chromosome_length

# Create a bar plot to visualize transcript density with rotated labels
barplot(chromosome_transcript_density$transcript_density,
        names.arg = chromosome_transcript_density$seqid,
        col = rainbow(nrow(chromosome_transcript_density)), # Assign colors
        xlab = "Chromosome",
        ylab = "Transcript Density",
        main = "Transcript Density on Each Chromosome",
        las = 2) # Rotate x-axis labels 90 degrees
```

## Transcript Density on Each Chromosome



```
# Identify chromosomes with min and max transcript density
min_transcript_density_chromosome <- chromosome_transcript_density$seqid[which.min(chromosome_transcript_density)]
max_transcript_density_chromosome <- chromosome_transcript_density$seqid[which.max(chromosome_transcript_density)]

cat("Chromosome with the min transcript density:", min_transcript_density_chromosome, "\n")
```

Chromosome with the min transcript density: chrY

```
cat("Chromosome with the max transcript density:", max_transcript_density_chromosome, "\n")
```

Chromosome with the max transcript density: chr19

## 4. Numbers and density of CDSs on each chromosome

(20%) Repeat 1 and 2 for CDS.

### 4.1 Repeat 1



```

# Filter GFF to include only chr1-22 and X,Y chromosomes
chromosomes_of_interest <- c("chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8", "chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18", "chr19", "chr20", "chr21", "chr22", "chrX", "chrY")
GFF_filtered <- GFF[GFF$seqid %in% chromosomes_of_interest, ]

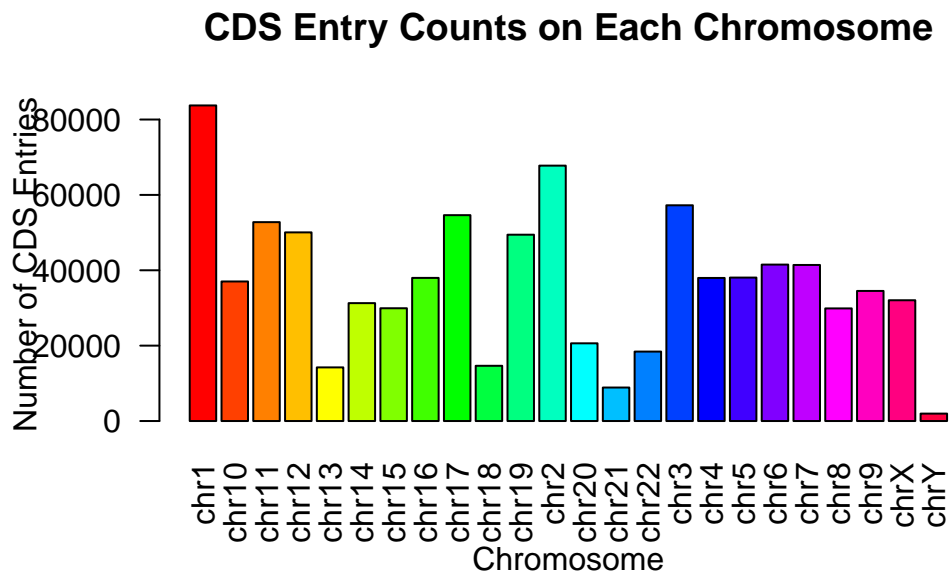
# Filter the data for CDS entries
cds_entries <- GFF_filtered[GFF_filtered$type == "CDS", ]

# Calculate the total number of CDS entries on each chromosome
chromosome_cds_counts <- table(cds_entries$seqid)

# Define colors for the bars (you can customize these colors)
bar_colors <- rainbow(length(chromosome_cds_counts))

# Create a bar plot to visualize CDS entry counts with colors
barplot(chromosome_cds_counts,
        names.arg = names(chromosome_cds_counts),
        xlab = "Chromosome",
        ylab = "Number of CDS Entries",
        main = "CDS Entry Counts on Each Chromosome",
        col = bar_colors,
        las = 2) # Specify the colors for the bars

```



```
# Calculate the chromosome with the minimum and maximum CDS entry counts
min_cds_chromosome <- names(chromosome_cds_counts)[which.min(chromosome_cds_counts)]
max_cds_chromosome <- names(chromosome_cds_counts)[which.max(chromosome_cds_counts)]

cat("Chromosome with the minimum CDS entry count:", min_cds_chromosome, "\n")
```

Chromosome with the minimum CDS entry count: chrY

```
cat("Chromosome with the maximum CDS entry count:", max_cds_chromosome, "\n")
```

Chromosome with the maximum CDS entry count: chr1

## 4.2 Repeat 2

```
# Filter GFF to include only chr1-22 and X,Y chromosomes
chromosomes_of_interest <- c("chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8", "chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18", "chr19", "chr20", "chr21", "chr22", "chrX", "chrY")
GFF_filtered <- GFF[GFF$seqid %in% chromosomes_of_interest, ]

# Filter the data for CDS entries
cds_entries <- GFF_filtered[GFF_filtered$type == "CDS", ]

# Calculate the total number of CDS entries on each chromosome
chromosome_cds_counts <- table(cds_entries$seqid)

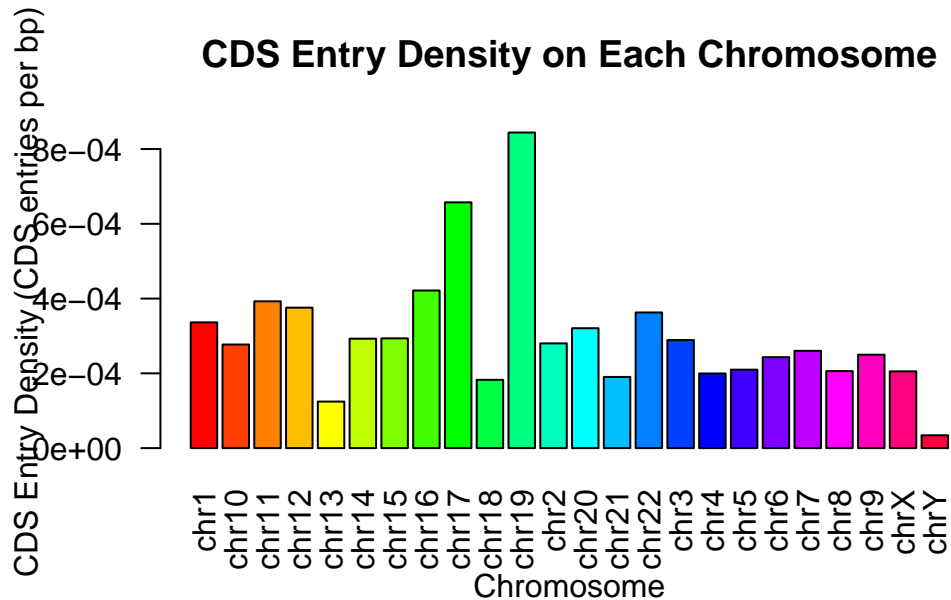
# Calculate chromosome lengths
chromosome_lengths <- aggregate(cds_entries$end, list(seqid = cds_entries$seqid), max)
colnames(chromosome_lengths) <- c("seqid", "chromosome_length")

# Calculate CDS entry density (CDS entries per base pair)
chromosome_cds_density <- merge(chromosome_lengths, data.frame(seqid = names(chromosome_cds_counts), chromosome_cds_counts))
chromosome_cds_density$cds_density <- chromosome_cds_density$num_cds / chromosome_cds_density$chromosome_length

# Define colors for the bars (you can customize these colors)
bar_colors <- rainbow(length(chromosome_cds_density$seqid))

# Create a bar plot to visualize CDS entry density with colors
barplot(chromosome_cds_density$cds_density,
        names.arg = chromosome_cds_density$seqid,
        xlab = "Chromosome",
```

```
ylab = "CDS Entry Density (CDS entries per bp)",
main = "CDS Entry Density on Each Chromosome",
col = bar_colors,
las = 2) # Specify the colors for the bars
```



```
# Calculate the chromosome with the minimum and maximum CDS entry density
min_cds_density_chromosome <- chromosome_cds_density$seqid[which.min(chromosome_cds_density)]
max_cds_density_chromosome <- chromosome_cds_density$seqid[which.max(chromosome_cds_density)]

cat("Chromosome with the minimum CDS entry density:", min_cds_density_chromosome, "\n")
```

Chromosome with the minimum CDS entry density: chrY

```
cat("Chromosome with the maximum CDS entry density:", max_cds_density_chromosome, "\n")
```

Chromosome with the maximum CDS entry density: chr19

## 5. Overall statistics

### (5%) 5.1 How many intervals are annotated in this version?

```
total_intervals <- nrow(GFF)
cat("Total number of annotated intervals:", total_intervals, "\n")
```

Total number of annotated intervals: 3422649

### (5%) 5.2 What is the total number of genes annotated in the human genome?

```
total_genes <- sum(GFF$type == "gene")
cat("Total number of genes in the human genome:", total_genes, "\n")
```

Total number of genes in the human genome: 62754

### (5%) 5.3 How many are protein coding genes?

```
pattern <- "gene_type=(^[^;]*)"
m <- regexec(pattern, GFF[, "attributes"])
gene_type <- sapply(
  regmatches(GFF[, "attributes"], m),
  function(e) {return(e[2])})
#
# Very slow alternative to extract gene names:
# d <- data.frame(regmatches(gff3[, "tag"], m))
# gene_names <- d[2, ]
GFF$gene_type2 = gene_type

total_protein_coding <- sum(GFF$gene_type2 == "protein_coding" & GFF$type == "gene")
cat("Total number of protein_coding in the human genome:", total_protein_coding, "\n")
```

Total number of protein\_coding in the human genome: 20070

### (5%) 5.4 How many are noncoding genes?

```
total_noncoding <- sum(GFF$gene_type2 != "protein_coding" & GFF$type == "gene")  
  
cat("Total number of noncoding in the human genome:", total_noncoding, "\n")
```

Total number of noncoding in the human genome: 42684