Module 1: Introduction to PheCAP data

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The goal of phenotyping is to predict patients' disease status from electronic health record data.

In this module, we will go through a public released dataset from an R package PheCAP to get hands-on experience of phenotyping.

```
# Load the packages.
packages <- c("tidyverse", "PheCAP", "corrplot", "ggplot2")

# Check if the pacakages are missing or not.
# If missing, install automatically.
# If not missing, load the package.
package.check <- lapply(
      packages,
      FUN = function(x) {
        if (!require(x, character.only = TRUE)) {
          install.packages(x, dependencies = TRUE)
          library(x, character.only = TRUE)
        }
      }
    )
```

PheCAP

https://celehs.github.io/PheCAP/

The most likely explanation is that this is a random sample of patients (for public release from a previous study) in the Partner's EHR database (4.6 million patients) with diabetes mellitus (DM) and who met

(i) an initial filter for CAD: $\geq$ 1 ICD9 code for CAD (410.x, 411.x, 412.x, 414.x, 413.x), or
(ii) $\geq$ 1 NLP mention for any CAD related concepts: CAD, CAD procedures, CAD biomarkers, positive stress test.

```
# Load helper functions.
source("../Rscripts/helper_function.R")
```

PheCAP data

```
data(ehr_data)
data <- PhecapData(ehr_data, "healthcare_utilization", "label", 0.4, patient_id = "patient_id")
data
```

```
## PheCAP Data
## Feature: 10000 observations of 587 variables
## Label: 119 yes, 62 no, 9819 missing
## Size of training samples: 109
## Size of validation samples: 72
```

What do you observe?

- 10,000 patients and 587 features.
- Label is subjective to missing.
- Split into training and validation set.

Elementary data exploration

```
ehr_data %>% head()
```

- Labels: "label", whether the patient has the disease, **extracted by clinicians' chart review**
- Features: "main_ICD", "main_NLP" refers to total number of billing codes or NLP mentions of the disease
- Features: "healthcare_utilization" refers to total number of notes the patient has
- Features: "CODx" (n = 10), "NLPx" (n = 574) refers to the counts of a specific code or NLP term, extracted by SQL or NLP

Missingness

```
colnames(ehr_data)[which( colMeans(is.na(ehr_data)) > 0)]
```

```
## [1] "label"
```

Only label is missing.

What is the prevalence of labels?

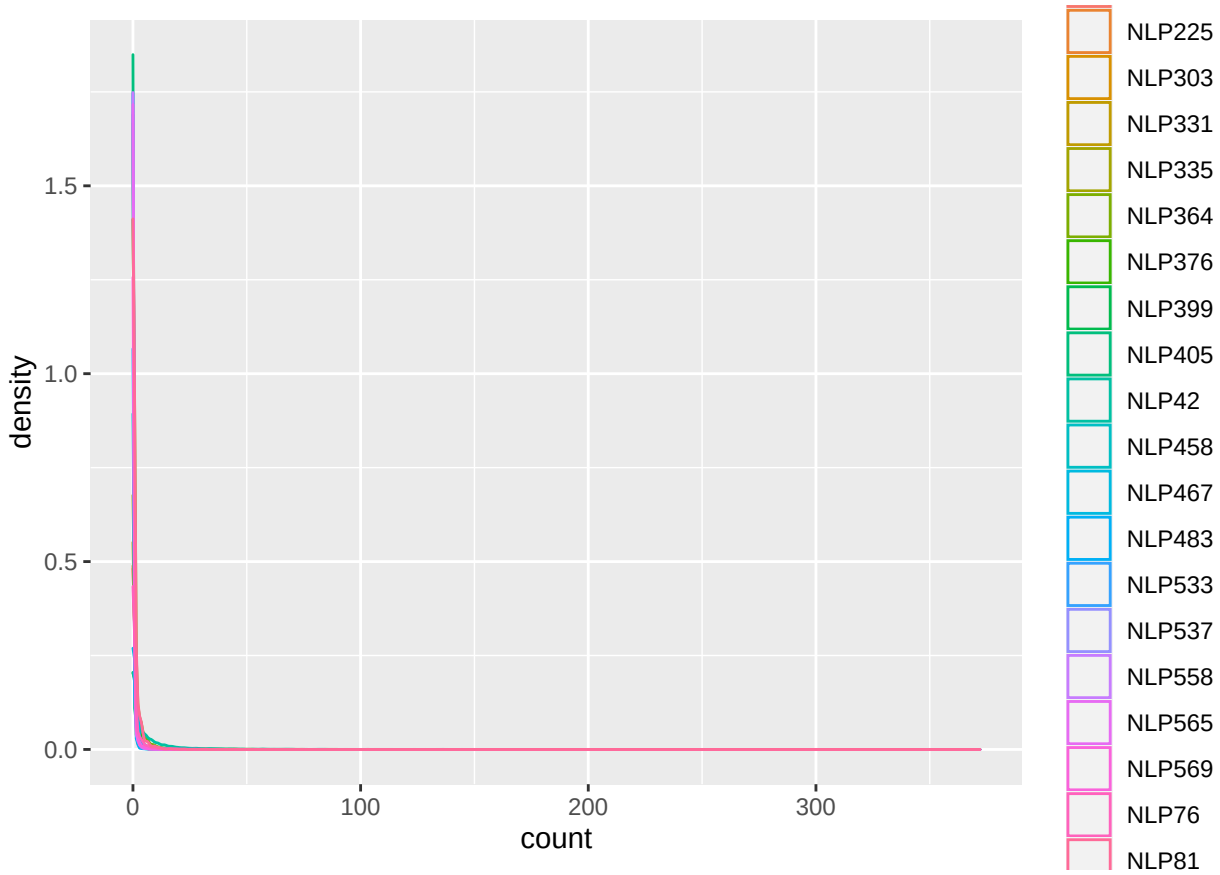
```
mean(ehr_data$label, na.rm = TRUE)
```

```
## [1] 0.6574586
```

How features are distributed?

- Let's randomly sample a few features first.
- Observe the densities.

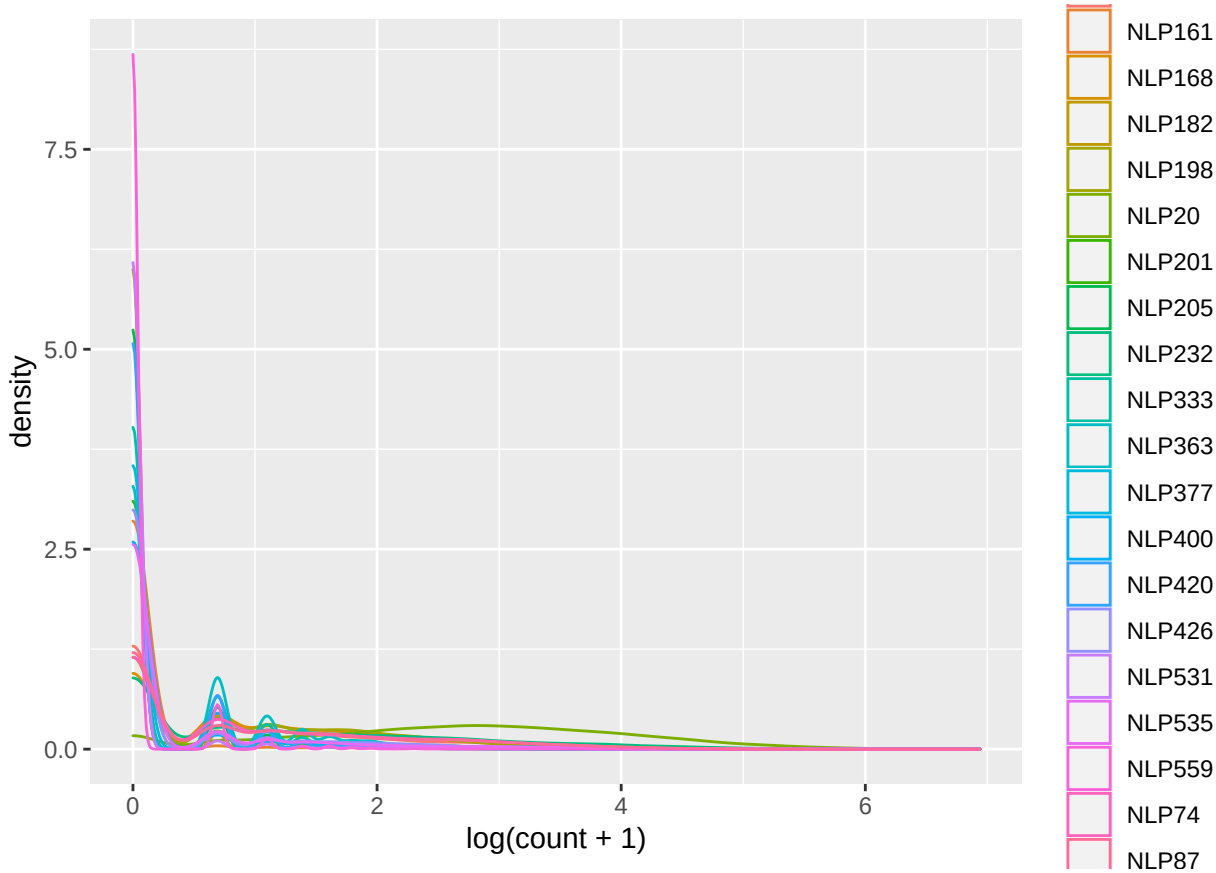
```
feature_index <- sample(c(3:ncol(ehr_data)), 20, replace = FALSE)
ehr_data[, feature_index] %>%
  pivot_longer(everything(), names_to = "feature", values_to = "count") %>%
  ggplot() +
  geom_density(aes(x = count, color = feature))
```



Too skewed? Use log transformation.

```
feature_index <- sample(c(3:ncol(ehr_data)), 20, replace = FALSE)

ehr_data[, feature_index] %>%
  pivot_longer(everything(), names_to = "feature", values_to = "count") %>%
  ggplot() +
  geom_density(aes(x = log(count + 1), color = feature))
```



Features are distributed very close to 0.

- Why using `log(count + 1)`?

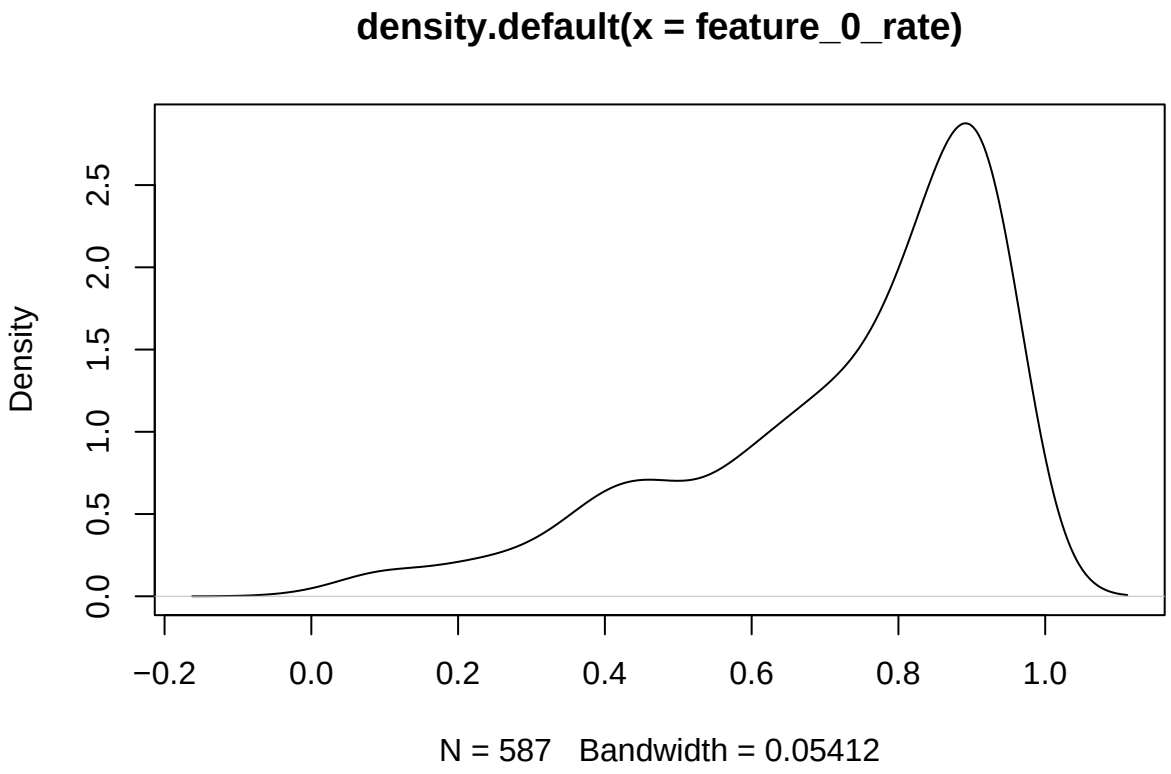
How many features are 0?

```
features <- ehr_data[, c(3:ncol(ehr_data))]
feature_0_rate <- colMeans(features == 0)

head(feature_0_rate, 10)
```

```
##               main_ICD               main_NLP healthcare_utilization
##                 0.5836                 0.6422                 0.0000
##                   COD1                   COD2                   COD3
##                 0.1649                 0.2297                 0.2580
##                   COD4                   COD5                   COD6
##                 0.6692                 0.4817                 0.2449
##                   COD7
##                 0.3476
```

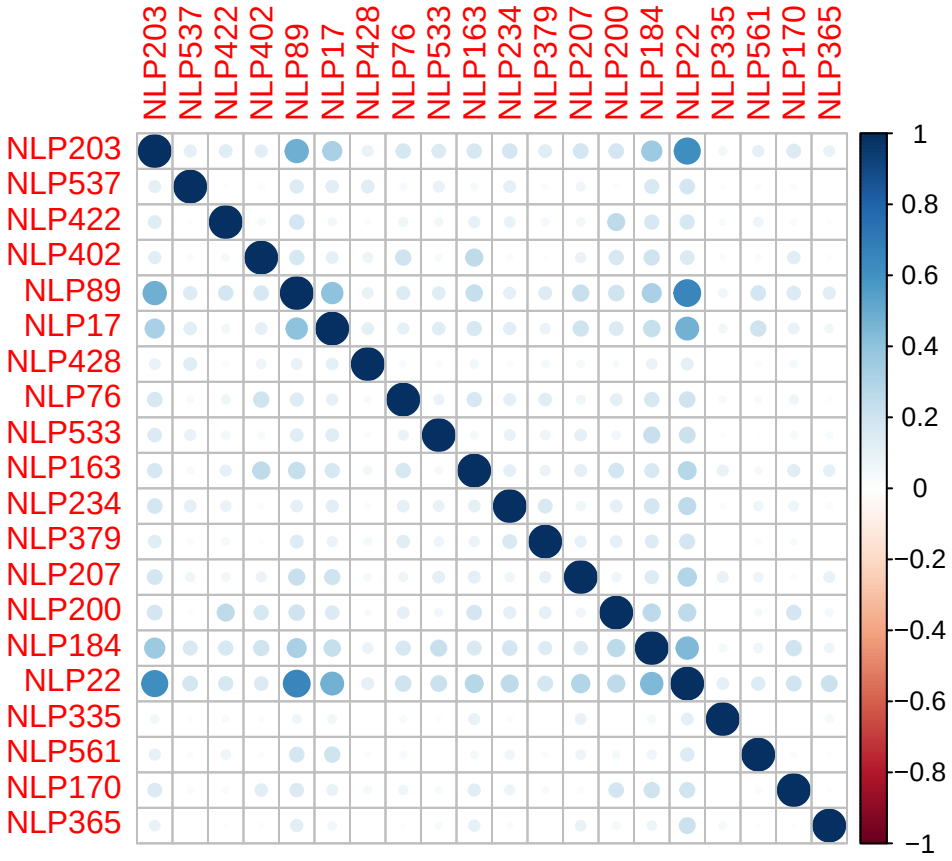
```
plot(density(feature_0_rate))
```



Most of the features have count 0 for most of the patients.

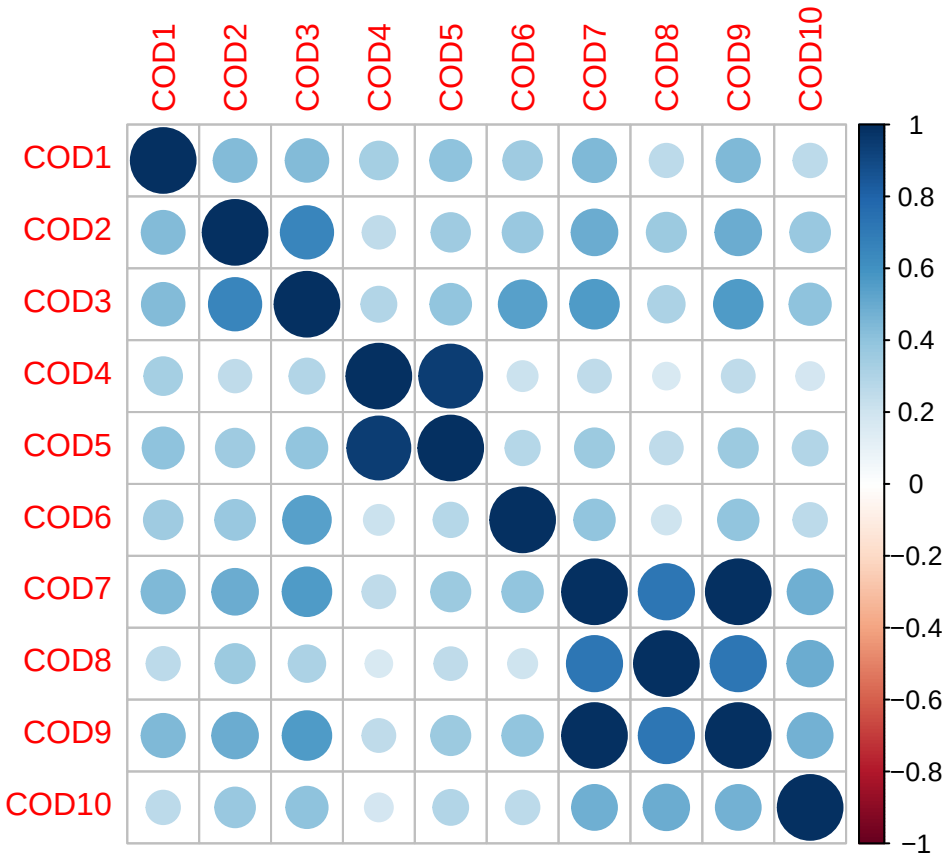
What are correlations between features?

```
feature_cor <- cor(features[feature_index])
corrplot::corrplot(feature_cor)
```



What about codified data?

```
feature_cor <- cor(features[4:13])
corrplot::corrplot(feature_cor)
```

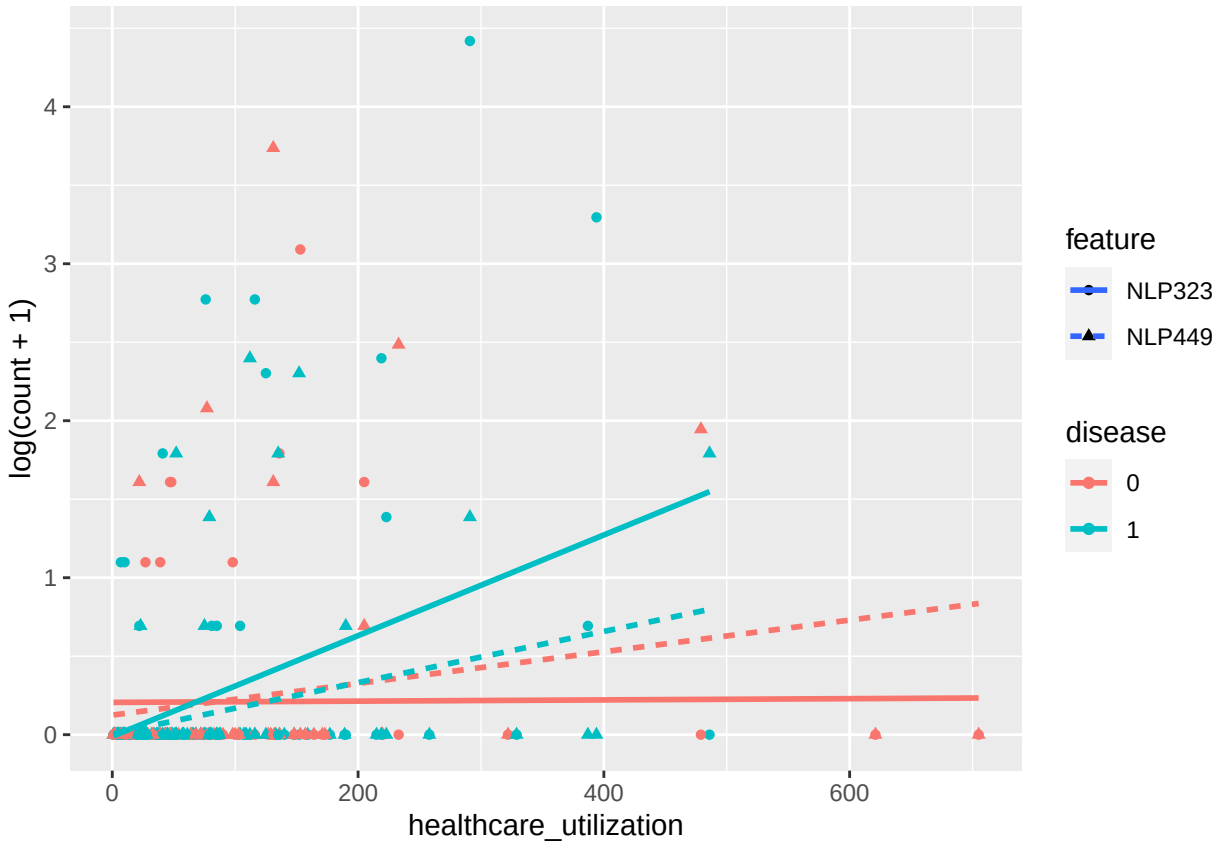


Healthcare utilization

"Healthcare_utilization" refers to total number of notes the patient has.

```
feature_index <- sample(c(3:ncol(ehr_data)), 2, replace = FALSE)

ehr_data %>%
  dplyr::select(label, healthcare_utilization, feature_index) %>%
  filter(!is.na(label)) %>%
  mutate(disease = factor(label)) %>%
  dplyr::select(-label) %>%
  pivot_longer(c(everything(), -disease, -healthcare_utilization),
               names_to = "feature", values_to = "count") %>%
  ggplot(aes(x = healthcare_utilization, y = log(count + 1), color = disease, shape = feature, linetype
  geom_point() +
  geom_smooth(method='lm', se = FALSE, size=1)
```



More healthcare utilization, more featuer counts; but not necessary the patient get the disease. Therefore, we need to adjust for the healthcare utilization when model fitting.