# 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 packages 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

```
load('../data/CAD_norm_pub.rda')
```

## Elementary data exploration

```
sum(!is.na(y))
```

```
## [1] 181
```

```
x %>% head()
```

- Labels: "y", whether the patient has the disease, **extracted by clinicians' chart review**
- Features: "surrogates" refers to total number of billing codes + 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

What do you observe?

- 4,164 patients and 586 features.
- Label is subjective to missing.

### What is the prevalence of labels?

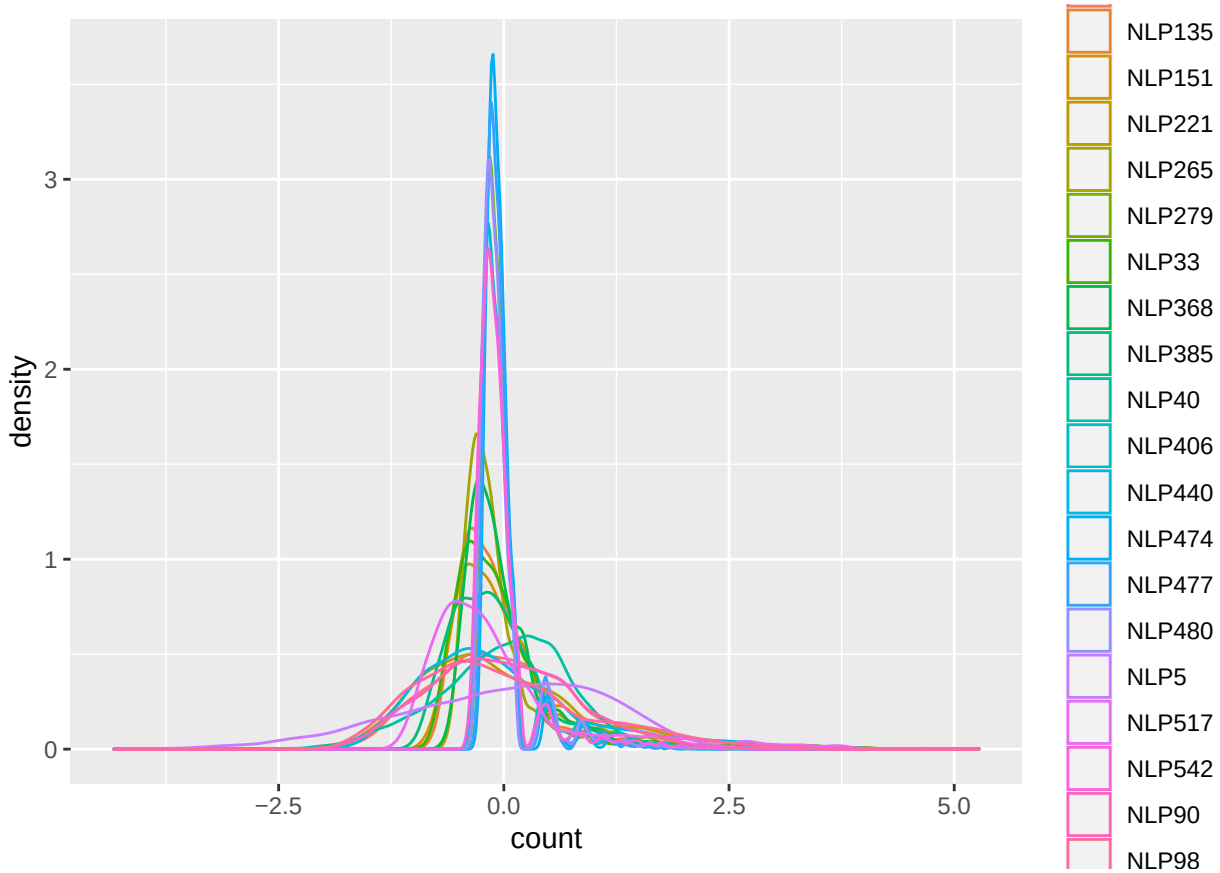
```
mean(y, na.rm = TRUE)
```

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

### How features are distributed?

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

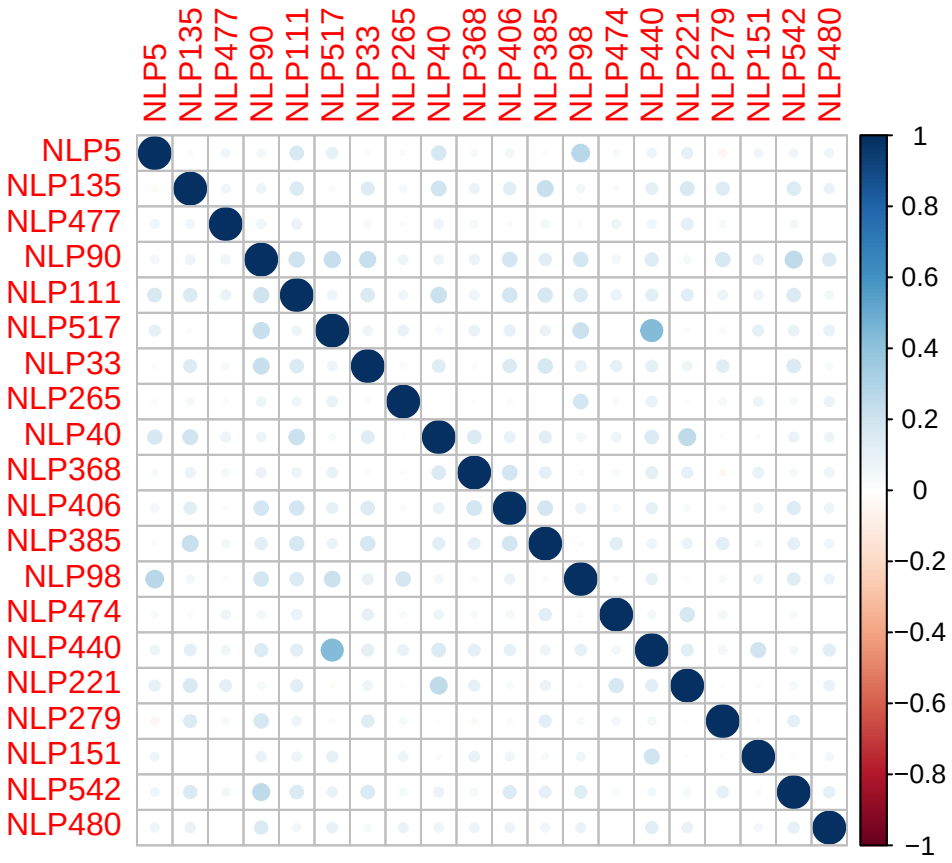
```
feature_index <- sample(c(1:ncol(x)), 20, replace = FALSE)

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




All the features are already orthogonalized and standardized.

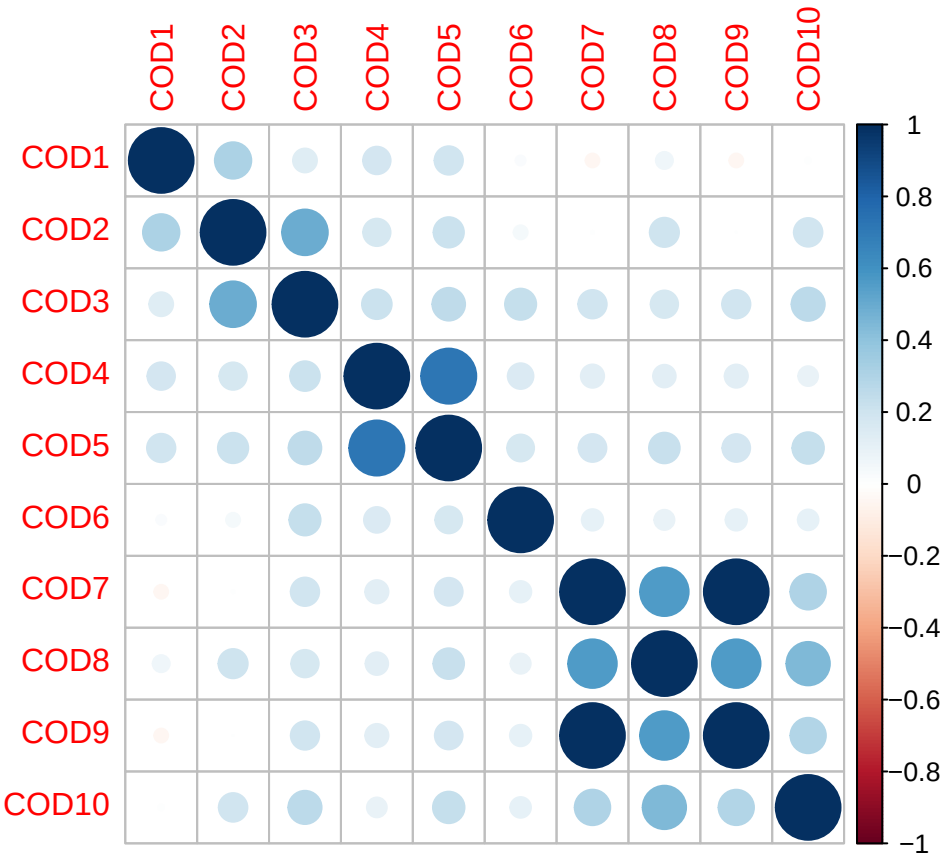
### What are correlations between features?

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




### What about codified data?

```
feature_cor <- cor(x[3:12])
corrplot::corrplot(feature_cor)
```


COD1
COD2
COD3
COD4
COD5
COD6
COD7
COD8
COD9
COD10
1
0.8
0.6
0.4
0.2
0
−0.2
−0.4
−0.6
−0.8
−1