Predicting Genetic Disorders

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Objective and Hypothesis

[. . .]

Data Load and Validation

```
# Load dataset(s)
gd_df <- read.csv("../data/train_genetic_disorders.csv", header = TRUE)

# Data validation and understanding, including structure, content, and statistical characteristics covered below
```

Data Structure Review

```
# Summarize base dataset and [optionally] sample rows
str(gd_df)
```

```
'data.frame':   22083 obs. of  45 variables:
 $ Patient.Id                                    : chr  "PID0x6418" "PID0x25d5" "PID0x4a82" "PID0x4ac8" ...
 $ Patient.Age                                   : int  2 4 6 12 11 14 3 3 11 4 ...
 $ Genes.in.mother.s.side                        : chr  "Yes" "Yes" "Yes" "Yes" ...
 $ Inherited.from.father                         : chr  "No" "Yes" "No" "No" ...
 $ Maternal.gene                                 : chr  "Yes" "No" "No" "Yes" ...
 $ Paternal.gene                                 : chr  "No" "No" "No" "No" ...
 $ Blood.cell.count..mcL.                        : num  4.76 4.91 4.89 4.71 4.72 ...
 $ Patient.First.Name                            : chr  "Richard" "Mike" "Kimberly" "Jeffery" ...
 $ Family.Name                                   : chr  "" "" "" "Hoelscher" ...
 $ Father.s.name                                 : chr  "Larre" "Brycen" "Nashon" "Aayaan" ...
 $ Mother.s.age                                  : int  NA NA 41 21 32 NA NA 40 45 44 ...
 $ Father.s.age                                  : int  NA 23 22 NA NA NA 63 NA 44 42 ...
 $ Institute.Name                                : chr  "Boston Specialty & Rehabilitation Hospital" "St. Margaret's Ho
spital For Women" "" "" ...
 $ Location.of.Institute                         : chr  "55 FRUIT ST\nCENTRAL, MA 02114\n(42.36247485742686, -71.069247
24545246)" "1515 COMMONWEALTH AV\nALLSTON/BRIGHTON, MA 02135\n(42.34665771451756, -71.14136122385321)" "-" "55 FRUIT ST\n
CENTRAL, MA 02114\n(42.36247485742686, -71.06924724545246)" ...
 $ Status                                        : chr  "Alive" "Deceased" "Alive" "Deceased" ...
 $ Respiratory.Rate..breaths.min.                : chr  "Normal (30-60)" "Tachypnea" "Normal (30-60)" "Tachypnea" ...
 $ Heart.Rate..rates.min                         : chr  "Normal" "Normal" "Tachycardia" "Normal" ...
 $ Test.1                                        : int  0 NA 0 0 0 0 NA 0 0 0 ...
 $ Test.2                                        : int  NA 0 0 0 0 0 0 0 0 0 ...
 $ Test.3                                        : int  NA 0 0 0 0 0 0 NA 0 0 ...
 $ Test.4                                        : int  1 1 1 1 1 1 1 1 1 1 ...
 $ Test.5                                        : int  0 0 0 0 0 0 0 0 0 0 ...
 $ Parental.consent                              : chr  "Yes" "Yes" "Yes" "Yes" ...
 $ Follow.up                                     : chr  "High" "High" "Low" "High" ...
 $ Gender                                        : chr  "" "" "" "Male" ...
 $ Birth.asphyxia                                : chr  "" "No" "No record" "Not available" ...
 $ Autopsy.shows.birth.defect..if.applicable.    : chr  "Not applicable" "None" "Not applicable" "No" ...
 $ Place.of.birth                                : chr  "Institute" "" "" "Institute" ...
 $ Folic.acid.details..peri.conceptional.        : chr  "No" "Yes" "Yes" "No" ...
 $ H.O.serious.maternal.illness                  : chr  "" "Yes" "No" "Yes" ...
 $ H.O.radiation.exposure..x.ray.                : chr  "No" "Not applicable" "Yes" "-" ...
 $ H.O.substance.abuse                           : chr  "No" "Not applicable" "" "Not applicable" ...
 $ Assisted.conception.IVF.ART                   : chr  "No" "No" "Yes" "" ...
 $ History.of.anomalies.in.previous.pregnancies  : chr  "Yes" "Yes" "Yes" "Yes" ...
 $ No..of.previous.abortion                      : int  NA NA 4 1 4 0 3 1 0 1 ...
 $ Birth.defects                                 : chr  "" "Multiple" "Singular" "Singular" ...
 $ White.Blood.cell.count..thousand.per.microliter.: num  9.86 5.52 NA 7.92 4.1 ...
 $ Blood.test.result                             : chr  "" "normal" "normal" "inconclusive" ...
 $ Symptom.1                                     : int  1 1 0 0 0 1 0 0 1 0 ...
 $ Symptom.2                                     : int  1 NA 1 0 0 0 0 0 1 0 ...
 $ Symptom.3                                     : int  1 1 1 1 0 0 0 1 1 1 ...
 $ Symptom.4                                     : int  1 1 1 0 0 1 0 NA 0 1 ...
 $ Symptom.5                                     : int  1 0 1 0 NA 0 0 0 1 1 ...
 $ Genetic.Disorder                              : chr  "Mitochondrial genetic inheritance disorders" "" "Multifactoria
l genetic inheritance disorders" "Mitochondrial genetic inheritance disorders" ...
 $ Disorder.Subclass                             : chr  "Leber's hereditary optic neuropathy" "Cystic fibrosis" "Diabet
```

```
es" "Leigh syndrome" ...
```

```
#head(gd_df, 3)
```

Preliminary Feature Reduction (clearly n/a to Objective and Hypothesis)

```
# Define n/a columns and subset dataframe; Note retaining "some" informational variables like "Institute.Name" for
#   possible descriptive analytic purposes
drop_cols <- c("Patient.Id",
               "Patient.First.Name",
               "Family.Name",
               "Father.s.name",
               "Institute.Name",
               "Location.of.Institute",
               "Status",
               "Test.1",
               "Test.2",
               "Test.3",
               "Test.4",
               "Test.5",
               "Parental.consent",
               "Place.of.birth")
gd_df <- gd_df[ , !(names(gd_df) %in% drop_cols)]
```

Class Target and Label Review

```
# Check for missing labels; set aside where missing
missing_target <- which(is.na(gd_df$Disorder.Subclass) | (gd_df$Disorder.Subclass == ""))
cat("Rows pre-subset for missing labels: ", format(nrow(gd_df), format = "d", big.mark = ","), sep = "")
```

```
Rows pre-subset for missing labels: 22,083
```

```
gd_hold_df <- gd_df[missing_target, ]
gd_df <- gd_df[-missing_target, ]
cat("Held rows with missing labels: ", format(nrow(gd_hold_df), format = "d", big.mark = ","), sep = "")
```

```
Held rows with missing labels: 3,140
```

```
cat("Net rows (labeled): ", format(nrow(gd_df), format = "d", big.mark = ","), sep = "")
```

```
Net rows (labeled): 18,943
```

```
# Show frequency distribution for [prospective] target class(es)
show_frequency <- function(desc, c) {
  t <- as.data.frame(prop.table(table(c)))
  colnames(t) <- c("Class", "Frequency")
  cat(desc, "\n"); print(t[order(-t$Freq, t$Class), 1:2], row.names = FALSE)
}
show_frequency("Pre-Split Frequency Distribution", gd_df$Disorder.Subclass)
```

```
Pre-Split Frequency Distribution
                             Class Frequency
                      Leigh syndrome     0.258
                Mitochondrial myopathy     0.222
                       Cystic fibrosis     0.173
                             Tay-Sachs     0.142
                              Diabetes     0.092
                       Hemochromatosis     0.068
 Leber's hereditary optic neuropathy     0.032
                            Alzheimer's     0.008
                                 Cancer     0.005
```

```
# Move the target class to "top" of dataframe so column removals don't impact
gd_df <- gd_df[ , c(ncol(gd_df), 1:(ncol(gd_df) - 1))]
target_col = 1



gd_df$Disorder.Subclass <- gsub("'", "", gd_df$Disorder.Subclass, fixed = TRUE)
gd_df$Disorder.Subclass <- gsub(" ", ".", gd_df$Disorder.Subclass, fixed = TRUE)
```

```
gd_df$Disorder.Subclass <- gsub("-", ".", gd_df$Disorder.Subclass, fixed = TRUE)
```

Data Splitting

```
# Split data 80/20 train/test, using caret's inherent stratified split to compensate for class imbalance
set.seed(1)
train_index <- createDataPartition(gd_df$Disorder.Subclass, times = 1, p = 0.80, list = FALSE)
train_df <- gd_df[train_index, ]
test_df <- gd_df[-train_index, ]
show_frequency("Post-Split Frequency Distribution (Train)", train_df$Disorder.Subclass)
```

```
Post-Split Frequency Distribution (Train)
                              Class Frequency
                      Leigh.syndrome     0.258
             Mitochondrial.myopathy     0.222
                    Cystic.fibrosis     0.173
                          Tay.Sachs     0.142
                           Diabetes     0.092
                    Hemochromatosis     0.068
 Lebers.hereditary.optic.neuropathy     0.032
                         Alzheimers     0.008
                             Cancer     0.005
```

Data Cleaning (and reduction)

Data (Sample) Characteristic Review for Pre-Processing

(Suppressing custom code for simplicity)

```
# Generate a summary (cursory) view of base dataset for initial understanding and pre-processing direction
univariate(train_df)
```

```
Summary Univariate Analysis (15,158 observations)
                     Type          NA BlankZ  Unique     Min     Max    Mean  Median Outlier< >Outlier Kurtosis Skewness
Disorder.Subclass    character                  9
Patient.Age          integer       6%     6%      15              14               7    No       Yes       0.017   -1.211
Genes.in.mother.s... character                  2
Inherited.from.fa... character             1%   3
Maternal.gene        character            12%   3
Paternal.gene        character                  2
Blood.cell.count.... numeric                 15,158   4.093   5.610   4.900   4.902    No       Yes      -0.011   -0.037
Mother.s.age         integer      26%           34      18      51              35    No       Yes      -0.006   -1.219
Father.s.age         integer      25%           45      20      64              42    No       Yes      -0.002   -1.210
Respiratory.Rate.... character             9%   3
Heart.Rate..rates... character             9%   3
Follow.up            character             9%   3
Gender               character             9%   4
Birth.asphyxia       character             9%   5
Autopsy.shows.bir... character             4%   5
Folic.acid.detail... character             9%   3
H.O.serious.mater... character             8%   3
H.O.radiation.exp... character             9%   5
H.O.substance.abuse  character             9%   5
Assisted.concepti... character             9%   3
History.of.anomal... character             9%   3
No..of.previous.a... integer       9%    18%   5               4               2    No       Yes       0.001   -1.292
Birth.defects        character             9%   3
White.Blood.cell.... numeric       9%          11,858   3.000  12.000   7.460   7.443    No       Yes       0.020   -0.979
Blood.test.result    character             9%   5
Symptom.1            integer       9%    37%   2               1               1    No       Yes      -0.369   -1.864
Symptom.2            integer       9%    40%   2               1               1    No       Yes      -0.197   -1.961
Symptom.3            integer       8%    41%   2               1               1    No       Yes      -0.166   -1.973
Symptom.4            integer       9%    45%   2               1                    No       Yes       0.010   -2.000
Symptom.5            integer       9%    48%   2               1                    No       Yes       0.146   -1.979
Genetic.Disorder     character             9%   4
```

Missing Values

```
# Genes.in.mother.s.side, Paternal.gene, Blood.cell.count..mcL., Status - n/a

# Impute basic integer values with medians
medianf <- function(x) {
```

```
 result <- median(x, na.rm = TRUE)
 if (is.integer(x))
   result <- as.integer(result)
 return(result)
}
median_cols = c("Patient.Age", "Mother.s.age", "Father.s.age", "No..of.previous.abortion")
for (n in median_cols) {
  train_df[n][is.na(train_df[n])] <- apply(train_df[n], 2, medianf)
  test_df[n][is.na(test_df[n])] <- apply(test_df[n], 2, medianf)
}

# Impute categorical blanks with common "notprovided"; note we could also impute these with categorical mode,
#   or most frequent categorical value of each column using the cmode() function below
cols_tofill <- c("Inherited.from.father",
                 "Maternal.gene",
                 "Respiratory.Rate..breaths.min.",
                 "Heart.Rate..rates.min",
                 "Follow.up",
                 "Gender",
                 "Birth.asphyxia",
                 "Autopsy.shows.birth.defect..if.applicable.",
                 "Folic.acid.details..peri.conceptional.",
                 "H.O.serious.maternal.illness",
                 "H.O.radiation.exposure..x.ray.",
                 "H.O.substance.abuse",
                 "Assisted.conception.IVF.ART",
                 "History.of.anomalies.in.previous.pregnancies",
                 "Birth.defects",
                 "Blood.test.result")
train_df[cols_tofill][train_df[cols_tofill] == ""] <- "notprovided"
test_df[cols_tofill][test_df[cols_tofill] == ""] <- "notprovided"

cmode <- function(x) {
  uniqx <- unique(na.omit(x))
  uniqx[which.max(tabulate(match(x, uniqx)))]
}

# Impute what appear to be masked "flag" columns iwth placeholder -1 values. . .
flag_cols <- c("Symptom.1", "Symptom.2", "Symptom.3", "Symptom.4", "Symptom.5")
train_df[flag_cols][is.na(train_df[flag_cols])] <- as.integer(-1)
test_df[flag_cols][is.na(test_df[flag_cols])] <- as.integer(-1)

# Impute mean for one numeric column
train_df$White.Blood.cell.count..thousand.per.microliter.[is.na(train_df$White.Blood.cell.count..thousand.per.microliter.
)] <-
  mean(train_df$White.Blood.cell.count..thousand.per.microliter., na.rm = TRUE)
test_df$White.Blood.cell.count..thousand.per.microliter.[is.na(test_df$White.Blood.cell.count..thousand.per.microliter.)]
 <-
  mean(test_df$White.Blood.cell.count..thousand.per.microliter., na.rm = TRUE)

# Note not using knnImpute for the limited number of numerical [prospective] features given that it
#   centers/scales, which is illogical for the values in this dataset
#pp <- preProcess(train_df[ , -target_col, drop = FALSE], method = "knnImpute", k = 10)
#train_df[ , -target_col] <- predict(pp, train_df[ , -target_col, drop = FALSE])
#test_df[ , -target_col] <- predict(pp, test_df[ , -target_col, drop = FALSE])

# Last on the list: Genetic.Disorder - we're not classifying to this but it is relevant/informational as a
#   superclass to the target Disorder.Subclass and shuold ultimately be imputed using similar Disorder.Subclass
#   observations which do have valid Genetic.Disorder values
```

Feature Updates (including variable types/formats, names)

```
# Re-type variables
factor_cols <- c("Genes.in.mother.s.side",
                 "Inherited.from.father",
                 "Maternal.gene",
                 "Paternal.gene",
                 "Respiratory.Rate..breaths.min.",
                 "Heart.Rate..rates.min",
                 "Follow.up",
                 "Gender",
                 "Birth.asphyxia",
                 "Autopsy.shows.birth.defect..if.applicable.",
                 "Folic.acid.details..peri.conceptional.",
                 "H.O.serious.maternal.illness",
                 "H.O.radiation.exposure..x.ray.",
                 "H.O.substance.abuse",
                 "Assisted.conception.IVF.ART",
                 "History.of.anomalies.in.previous.pregnancies",
                 "Birth.defects",
                 "Blood.test.result",
                 "Disorder.Subclass")
```

```
train_df[factor_cols] <- lapply(train_df[factor_cols], factor)
test_df[factor_cols] <- lapply(test_df[factor_cols], factor)
# Note dummy variables may be introduced below for e.g., logistic regression

# Simplify variable naming
# [TBD]

# Generate updated summary of base dataset
univariate(train_df)
```

```
Summary Univariate Analysis (15,158 observations)
                     Type          NA BlankZ  Unique     Min     Max    Mean  Median Outlier< >Outlier Kurtosis Skewness
Disorder.Subclass    factor                      9
Patient.Age          integer               6%       15              14               7    No       Yes       0.016   -1.090
Genes.in.mother.s... factor                      2
Inherited.from.fa... factor                      3
Maternal.gene        factor                      3
Paternal.gene        factor                      2
Blood.cell.count.... numeric                15,158   4.093   5.610   4.900   4.902    No       Yes      -0.011   -0.037
Mother.s.age         integer                    34      18      51              35    No       Yes      -0.048   -0.593
Father.s.age         integer                    45      20      64              42    No       Yes      -0.007   -0.600
Respiratory.Rate.... factor                      3
Heart.Rate..rates... factor                      3
Follow.up            factor                      3
Gender               factor                      4
Birth.asphyxia       factor                      5
Autopsy.shows.bir... factor                      5
Folic.acid.detail... factor                      3
H.O.serious.mater... factor                      3
H.O.radiation.exp... factor                      5
H.O.substance.abuse  factor                      5
Assisted.concepti... factor                      3
History.of.anomal... factor                      3
No..of.previous.a... integer              18%       5               4               2    No       Yes               -1.116
Birth.defects        factor                      3
White.Blood.cell.... numeric                11,859   3.000  12.000   7.460   7.460    No       Yes       0.021   -0.768
Blood.test.result    factor                      5
Symptom.1            integer              37%       3      -1       1               1    No       Yes      -0.769   -0.496
Symptom.2            integer              40%       3      -1       1                    No       Yes      -0.643   -0.624
Symptom.3            integer              41%       3      -1       1                    No       Yes      -0.626   -0.613
Symptom.4            integer              45%       3      -1       1                    No       Yes      -0.502   -0.679
Symptom.5            integer              48%       3      -1       1                    No       Yes      -0.413   -0.702
Genetic.Disorder     character             9%       4
```

Zero/Near-Zero Variances

```
# n/a for this dataset
```

Duplicate Values

```
# n/a for this dataset
```

“Noisy” Data

```
# n/a for this dataset
```

Data Transformation

Centering/Scaling (standardizing/normalizing)

```
# n/a for this dataset?
```

Statistical Characteristics (including distribution, skewness, outliers)

```
#summary(train_df)
```

Other Feature Engineering (transformation, aggregation, enrichment)

```
# n/a for this dataset?
```

Multivariate Analysis (and reduction)

Collinearity and Dependencies

```
# Calculate Cramer's V "measure of association" between nominal factor variables (uses Chi-square statistic)
cscorr <- PairApply(train_df[ , sapply(train_df, is.factor)], CramerV, symmetric = TRUE)

# Shorten variable names for ease of reviewing output matrix
rn <- rownames(cscorr)
for (n in 1:length(rownames(cscorr))) {
  rn[n] <- paste(rownames(cscorr)[n], " (", AscToChar(64 + n), ")", sep = "")
  rownames(cscorr)[n] <- paste(AscToChar(64 + n))
}
for (n in 1:length(colnames(cscorr)))
  colnames(cscorr)[n] <- paste(AscToChar(64 + n))

# Show master list of variable names along with output ("correlation") matrix
cat(rn, sep = "\n")
```

```
Disorder.Subclass (A)
Genes.in.mother.s.side (B)
Inherited.from.father (C)
Maternal.gene (D)
Paternal.gene (E)
Respiratory.Rate..breaths.min. (F)
Heart.Rate..rates.min (G)
Follow.up (H)
Gender (I)
Birth.asphyxia (J)
Autopsy.shows.birth.defect..if.applicable. (K)
Folic.acid.details..peri.conceptional. (L)
H.O.serious.maternal.illness (M)
H.O.radiation.exposure..x.ray. (N)
H.O.substance.abuse (O)
Assisted.conception.IVF.ART (P)
History.of.anomalies.in.previous.pregnancies (Q)
Birth.defects (R)
Blood.test.result (S)
```

```
cscorr
```

```
     A     B     C     D     E     F     G    H    I     J    K     L     M     N    O     P     Q     R    S
A 1.00 0.198 0.131 0.123 0.168 0.019 0.026 0.02 0.02 0.022 0.02 0.020 0.019 0.024 0.02 0.019 0.026 0.025 0.03
B 0.20 1.000 0.005 0.097 0.012 0.005 0.005 0.01 0.01 0.008 0.01 0.013 0.009 0.016 0.01 0.003 0.017 0.008 0.01
C 0.13 0.005 1.000 0.013 0.093 0.018 0.020 0.01 0.02 0.022 0.02 0.021 0.013 0.030 0.02 0.013 0.018 0.016 0.02
D 0.12 0.097 0.013 1.000 0.008 0.048 0.040 0.05 0.05 0.054 0.04 0.053 0.048 0.052 0.04 0.055 0.047 0.044 0.05
E 0.17 0.012 0.093 0.008 1.000 0.003 0.009 0.01 0.01 0.023 0.02 0.003 0.001 0.008 0.02 0.003 0.008 0.006 0.02
F 0.02 0.005 0.018 0.048 0.003 1.000 0.045 0.03 0.05 0.036 0.02 0.043 0.028 0.030 0.04 0.035 0.036 0.042 0.04
G 0.03 0.005 0.020 0.040 0.009 0.045 1.000 0.04 0.05 0.034 0.02 0.035 0.029 0.047 0.03 0.055 0.042 0.041 0.05
H 0.02 0.015 0.012 0.046 0.011 0.029 0.040 1.00 0.04 0.033 0.04 0.041 0.043 0.032 0.04 0.043 0.051 0.038 0.05
I 0.02 0.010 0.023 0.047 0.010 0.054 0.045 0.04 1.00 0.042 0.02 0.032 0.051 0.045 0.04 0.035 0.028 0.036 0.04
J 0.02 0.008 0.022 0.054 0.023 0.036 0.034 0.03 0.04 1.000 0.03 0.035 0.026 0.020 0.03 0.047 0.048 0.036 0.03
K 0.02 0.010 0.016 0.035 0.025 0.019 0.023 0.04 0.02 0.025 1.00 0.030 0.022 0.028 0.03 0.021 0.024 0.029 0.03
L 0.02 0.013 0.021 0.053 0.003 0.043 0.035 0.04 0.03 0.035 0.03 1.000 0.020 0.049 0.04 0.028 0.032 0.030 0.04
M 0.02 0.009 0.013 0.048 0.001 0.028 0.029 0.04 0.05 0.026 0.02 0.020 1.000 0.048 0.04 0.043 0.042 0.032 0.04
N 0.02 0.016 0.030 0.052 0.008 0.030 0.047 0.03 0.04 0.020 0.03 0.049 0.048 1.000 0.03 0.046 0.047 0.052 0.04
O 0.02 0.011 0.015 0.043 0.016 0.035 0.032 0.04 0.04 0.032 0.03 0.042 0.037 0.026 1.00 0.033 0.050 0.049 0.03
P 0.02 0.003 0.013 0.055 0.003 0.035 0.055 0.04 0.03 0.047 0.02 0.028 0.043 0.046 0.03 1.000 0.035 0.032 0.03
Q 0.03 0.017 0.018 0.047 0.008 0.036 0.042 0.05 0.03 0.048 0.02 0.032 0.042 0.047 0.05 0.035 1.000 0.032 0.04
R 0.02 0.008 0.016 0.044 0.006 0.042 0.041 0.04 0.04 0.036 0.03 0.030 0.032 0.052 0.05 0.032 0.032 1.000 0.04
S 0.03 0.013 0.018 0.052 0.016 0.036 0.046 0.05 0.04 0.029 0.03 0.042 0.041 0.037 0.03 0.031 0.041 0.044 1.00
```

Predictor Transformations (e.g., PCA)

Modeling

Feature Selection

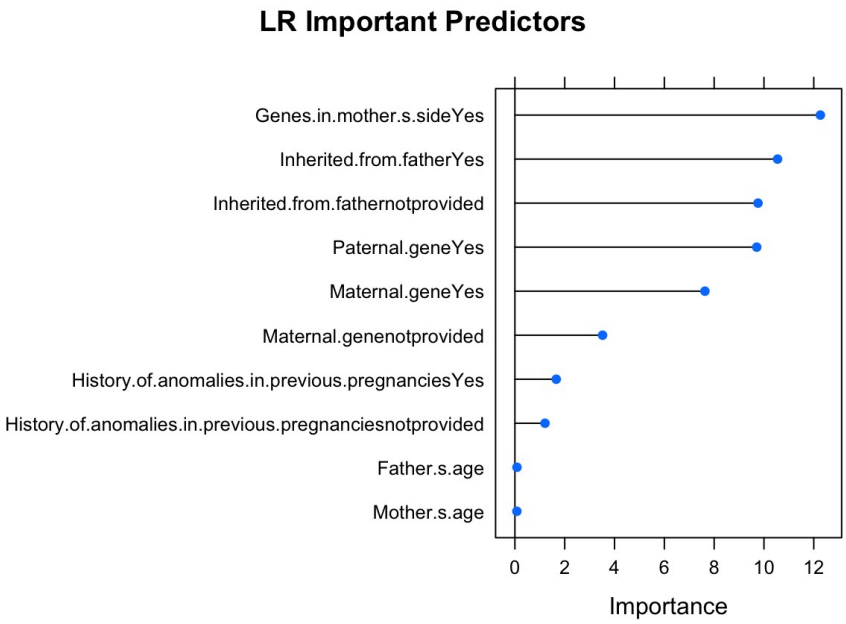
Training, Testing (validating), and Evaluation (iteration n)

```
# Create common control for models
set.seed(1)
fit_control <- trainControl(method = "cv",
                            number = 10,
                            savePredictions = "all",
                            classProbs = TRUE,
                            summaryFunction = multiClassSummary)

# Select predictors (features) for lr model
predictor_cols <- c("Genes.in.mother.s.side",
                    "Inherited.from.father",
                    "Maternal.gene",
                    "Paternal.gene",
                    "Mother.s.age",
                    "Father.s.age",
                    "History.of.anomalies.in.previous.pregnancies")
#*Genes.in.mother.s.side
#*Inherited.from.father
#*Maternal.gene
#*Paternal.gene
#Blood.cell.count..mcL.
#*Mother.s.age
#*Father.s.age
#Respiratory.Rate..breaths.min.
#Heart.Rate..rates.min
#Follow.up
#Gender
#Birth.asphyxia
#Autopsy.shows.birth.defect..if.applicable.
#Folic.acid.details..peri.conceptional.
#H.O.serious.maternal.illness
#H.O.radiation.exposure..x.ray.
#H.O.substance.abuse
#Assisted.conception.IVF.ART
#*History.of.anomalies.in.previous.pregnancies
#No..of.previous.abortion
#Birth.defects
#White.Blood.cell.count..thousand.per.microliter.
#Blood.test.result
#Symptom.1
#Symptom.2
#Symptom.3
#Symptom.4
#Symptom.5

# Train logistic regression model
invisible(capture.output(
  lr_fit <- train(x = train_df[ , predictor_cols, drop = FALSE],
                  y = train_df$Disorder.Subclass,
                  method = "multinom",
                  metric = "ROC",
                  trControl = fit_control)
))
#lr_fit
#lr_fit$finalModel

# Generate confusion matrix
lr_cm <- confusionMatrix(lr_fit, norm = "none")

# Simplify class names for more coherent confusion matrix, and output
for (n in 1:length(rownames(lr_cm$tab)))
  rownames(lr_cm$table)[n] <- paste(rownames(lr_cm$table)[n], " (", AscToChar(64 + n), ")", sep = "")
for (n in 1:length(colnames(lr_cm$tab)))
  colnames(lr_cm$table)[n] <- paste("Class", AscToChar(64 + n))
lr_cm
```

```
Cross-Validated (10 fold) Confusion Matrix

(entries are un-normalized aggregated counts)

                                      Reference
Prediction                             Class A Class B Class C Class D Class E Class F Class G Class H Class I
  Alzheimers (A)                             0       0       0       0       0       0       0       0       0
  Cancer (B)                                 0       0       0       0       0       0       0       0       0
  Cystic.fibrosis (C)                       43       0     364     236      25     116     326     169      88
  Diabetes (D)                               0       0       0       0       0       0       0       0       0
  Hemochromatosis (E)                        0       0       0       0       0       0       0       0       0
  Lebers.hereditary.optic.neuropathy (F)     0       0       0       0       0       0       0       0       0
  Leigh.syndrome (G)                        71      24    1842    1000     553     331    2632    2153    1256
  Mitochondrial.myopathy (H)                 5      50     414     159     453      39     954    1039     802
```

```
  Tay.Sachs (I)                              0        0        2        0        2        0        3        1        6

 Accuracy (average) : 0.2666
```

```
# Plot ROC curve
#lr_roc <- roc(response = lr_fit$pred$obs,
#              predictor = lr_fit$pred$Yes,
#              levels = rev(levels(lr_fit$pred$obs)))
#if (show_output) { plot(lr_roc, legacy.axes = TRUE, main = "LR ROC Curve",
#                        xlab = "Fall Positive Rate (Specificity)",
#                        ylab = "True Positive Rate (Sensitivity)") }
#if (show_output) { lr_roc$auc }

# Check for most important predictors
plot(varImp(lr_fit, scale = FALSE), top = 10, main = "LR Important Predictors")
```




```
varImp(lr_fit)
```

```
multinom variable importance

                                                 Overall
Genes.in.mother.s.sideYes                       100.0000
Inherited.from.fatherYes                         85.8991
Inherited.from.fathernotprovided                 79.4552
Paternal.geneYes                                 79.0347
Maternal.geneYes                                 61.9656
Maternal.genenotprovided                         28.2418
History.of.anomalies.in.previous.pregnanciesYes  12.9769
History.of.anomalies.in.previous.pregnanciesnotprovided  9.2783
Father.s.age                                      0.0418
Mother.s.age                                      0.0000
```

```
# Establish test_results dataframe
test_results <- data.frame(obs = test_df$Disorder.Subclass,
                           model_2_tbd = 0.0,
                           model_3_tbd = 0.0,
                           model_4_tbd = 0.0,
                           model_5_tbd = 0.0)

# Validate model vs. test data
test_results$lr <- predict(lr_fit, test_df[ , predictor_cols, drop = FALSE])
```

Optimization, Tuning, Selection

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