Patient Diagnosis with Machine Learning

Caitlin Ruble

July 2022

Introduction

Problem:

For a sick patient, the pathway to feeling better begins with an accurate diagnosis. Doctors are the first line responders to assess a patient's symptoms, categorize the patient with a diagnosis, and then create a treatment plan based on that diagnosis. While this approach can, and often does work, and has been the approach for the entirety of the history of medicine, it does require doctors to hold vast amounts of memorized knowledge, doesn't account for emerging diseases, and is at the mercy of human error. When ailments are misdiagnosed, patients suffer and cannot get the treatment they need. At best, they suffer for longer than they need to, and at worst their ailments can lead to permanent disabilities and even death. What if we could leverage all the canonical knowledge of the field of medicine and ask a machine to hold on to that for us, instead of relying on individual doctors to memorize and apply their knowledge perfectly? If we could build a machine learning model that correctly classified a patients diagnosis based on their presenting symptoms, we could save lives and reduce suffering by ensuring timely, accurate diagnoses, thus leading to timely, appropriate treatment. This is the problem we sought to solve!

Client:

Such a classifier would add monetized value to many entities across the medical industry. Patients who receive an accurate diagnosis and appropriate treatment the first time around will avoid unnecessary medical charges related to treating a misdiagnosis and are likelier to stay out of emergency situations related to their ailment. This equates to significant cost savings for hospital systems, health insurance providers, and other medical provider agencies, all while delivering better outcomes for patients.

Dataset:

The "Disease Prediction Using Machine Learning" dataset on Kaggle contains symptom data for 4920 individual observations. There are 132 binary symptom features, and a "prognosis" feature which holds the disease classification label.

Approach:

I will use several supervised machine learning techniques and select the best-performing one to build a classifier that can accurately classify a patient's diagnosis from the set of diseases included in the dataset, based on the presence/absence of the included symptoms, with >95% accuracy.

Data Cleaning

Description of Features:

The following is a complete list and description of the features included in the dataset:

File: Training.csv

prognosis: the categorical assignment of a disease label for each observation, our target. The 41 diagnoses included in the data set are listed in Table 1, below.

Unnamed: 133: an extra column containing all NaN values

symptom features: 132 binary feature columns, each containing a 0 to indicate absence and a 1 to indicate presence of the symptom for each observation. The individual feature names are listed in Table 2, below.

Table 1: List of the 41 disease labels included in the original data set under feature name "prognosis"

AIDS	Drug Reaction	Hypoglycemia
Acne	Fungal infection	Hypothyroidism
Alcoholic hepatitis	GERD	Osteoarthristis
Allergy	Gastroenteritis	Paralysis (brain hemorrhage)
Arthritis	Heart attack	Peptic ulcer diseae
Bronchial Asthma	hepatitis A	Pneumonia
Cervical spondylosis	Hepatitis B	Psoriasis
Chicken pox	Hepatitis C	Tuberculosis
Chronic cholestasis	Hepatitis D	Typhoid
Common Cold	Hepatitis E	Urinary tract infection
Dengue	Hypertension	Varicose veins
Diabetes	Hyperthyroidism	(vertigo) Paroymsal Positional Vertigo

Data Handling:

The .csv file was loaded into a Jupyter notebook as a pandas DataFrame. All columns were checked for the correct data type, and no changes to the data types were necessary.

Handling of Missing Values:

1. There was an extra feature column, 'Unnamed: 133,' which was found to contain all NaN values. This column was dropped from the data set.
2. The symptom feature 'fluid_overload' was found to contain only 0s for the entire data set, indicating it was adding no information. Additionally, there was a redundant symptom feature named 'fluid_overload.1' which *did* contain binary information. The original 'fluid_overload' column was deleted and the 'fluid_overload.1' column was renamed to 'fluid_overload' for ease of interpretation. This brought our symptom feature count to 131.

Handling of Formatting and Misspelling Issues:

Based on domain knowledge and careful review of the prognosis categories, several inconsistencies that could present interpretation issues in a production environment were identified and remedied. These formatting changes were carried out on the 'prognosis' feature cells, and are summarized in Table 3.

Table 2: List of binary symptom feature columns in the original data set		
abdominal_pain	foul_smell_of urine	receiving_blood_transfusion
abnormal_menstruation	headache	receiving_unsterile_injections
acidity	high_fever	red_sore_around_nose
acute_liver_failure	hip_joint_pain	red_spots_over_body
altered_sensorium	history_of_alcohol_consumption	redness_of_eyes
anxiety	increased_appetite	restlessness
back_pain	indigestion	runny_nose
belly_pain	inflammatory_nails	rusty_sputum
blackheads	internal_itching	scurring
bladder_discomfort	irregular_sugar_level	shivering
blister	irritability	silver_like_dusting
blood_in_sputum	irritation_in_anus	sinus_pressure
bloody_stool	itching	skin_peeling
blurred_and_distorted_vision	joint_pain	skin_rash
breathlessness	knee_pain	slurred_speech
brittle_nails	lack_of_concentration	small_dents_in_nails
bruising	lethargy	spinning_movements
burning_micturition	loss_of_appetite	spotting_ urination
chest_pain	loss_of_balance	stiff_neck
chills	loss_of_smell	stomach_bleeding
cold_hands_and_feets	malaise	stomach_pain
coma	mild_fever	sunken_eyes
congestion	mood_swings	sweating
constipation	movement_stiffness	swelled_lymph_nodes
continuous_feel_of_urine	mucoid_sputum	swelling_joints
continuous_sneezing	muscle_pain	swelling_of_stomach
cough	muscle_wasting	swollen_blood_vessels
cramps	muscle_weakness	swollen_extremeties
dark_urine	nausea	swollen_legs
dehydration	neck_pain	throat_irritation
depression	nodal_skin_eruptions	toxic_look_(typhos)
diarrhoea	obesity	ulcers_on_tongue
dischromic _patches	pain_behind_the_eyes	unsteadiness
distention_of_abdomen	pain_during_bowel_movements	visual_disturbances
dizziness	pain_in_anal_region	vomiting
drying_and_tingling_lips	painful_walking	watering_from_eyes
enlarged_thyroid	palpitations	weakness_in_limbs
excessive_hunger	passage_of_gases	weakness_of_one_body_side
extra_marital_contacts	patches_in_throat	weight_gain
family_history	phlegm	weight_loss
fast_heart_rate	polyuria	yellow_crust_ooze
fatigue	prominent_veins_on_calf	yellow_urine
fluid_overload	puffy_face_and_eyes	yellowing_of_eyes
fluid_overload.1	pus_filled_pimples	yellowish_skin

Table 3: Formatting changes to disease classifications in ‘prognosis’ feature	
Original	**Cleaned**
‘hepatitis A’	‘Hepatitis A’
‘Osteoarthristis’	‘Osteoarthritis’
‘Dimorphic hemmorhoids(piles)’	‘Dimorphic hemorrhoids (piles)’
‘(vertigo) Paroymsal Positional Vertigo’	‘Paroxysmal positional vertigo’
‘Peptic ulcer diseae’	‘Peptic ulcer disease’

Exploratory Data Analysis

Distributions of Symptoms for Each Disease:

Frequency plots of each symptom assigned to each disease were plotted, revealing that each disease category has a distinct combination of symptoms associated with it, creating a visually distinct "signature." We know that each disease can have up to 120 counts of each symptom. When a frequency bar reached 120 (the top of the plot), it indicated that every single instance of that disease in the training data was positive for that symptom. While this isn't exactly rare (check out the "Common cold" or "Hyperthyroidism" charts, among others, for examples), it was far more common in our data set for many, but not all, of the instances of a disease to show a particular symptom. No symptom frequency bar was less than ~100 instances out of 120. Each prognosis had multiple symptoms associated with it. Several demonstrative examples are reproduced in Figure 1, below.

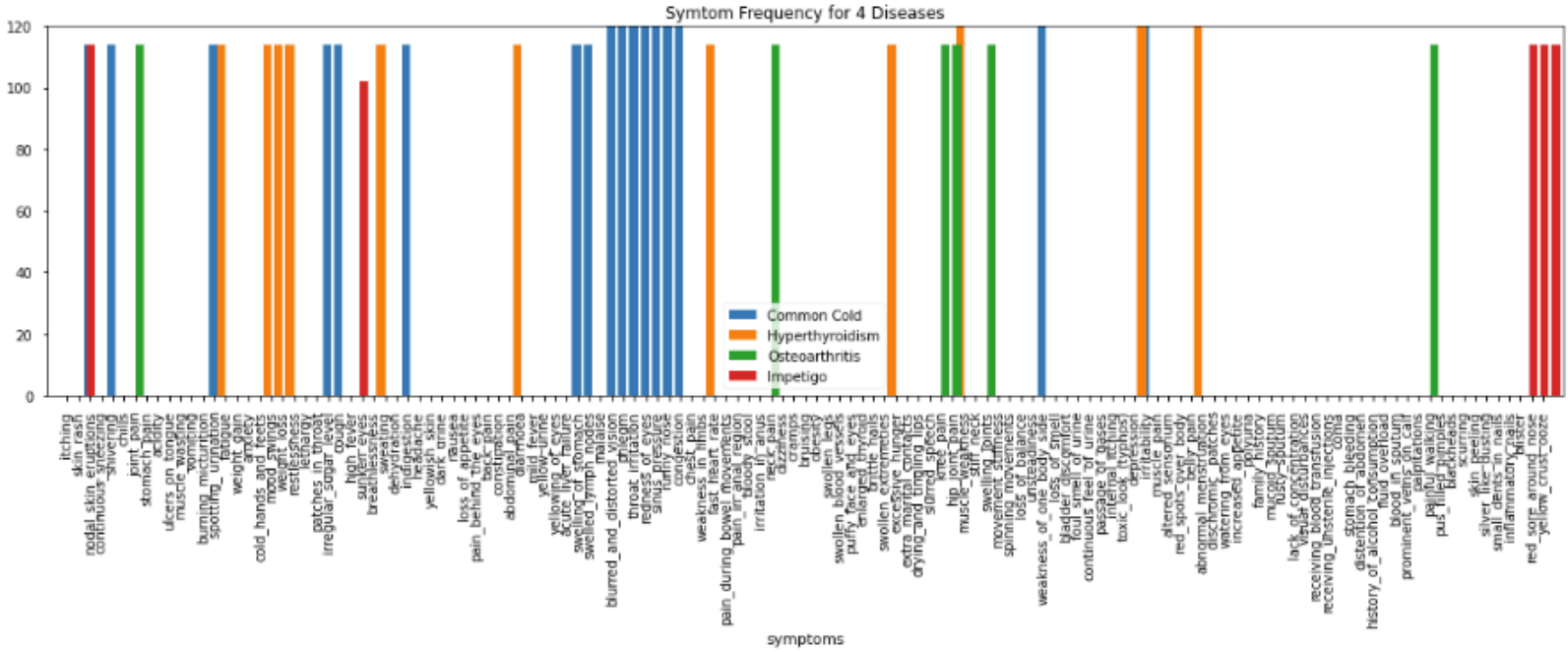

Figure 1: Symptom Frequency Chart for 4 Diseases: Common Cold, Hyperthyroidism, Osteoarthritis, Impetigo. These are meant to be demonstrative of the different symptom "signatures" of each disease in the dataset showed, and how while some symptoms are implicated in 100% of the cases of a disease, many are not.

Distributions of Diseases Associated with Each Symptom:

Frequency plots of which diseases were associated with each symptom were also plotted. Figure 2 shows a representative sample of 4 of the symptom features and which diseases were associated with them at which frequencies. There are several conclusions we drew:

1. Some symptoms were only indicated in 1 disease prognosis. (See 'yellow_crust_ooze')
2. Some symptoms were only indicated in 1 disease prognosis *and* are indicated in every instance of that prognosis in the training set. (See 'palpitations')
3. Some symptoms were indicated in more than one prognosis. (see 'weight_loss')
4. Some symptoms were indicated in more than one prognosis and were always indicated in one or more of the possibilities. (see 'malaise')

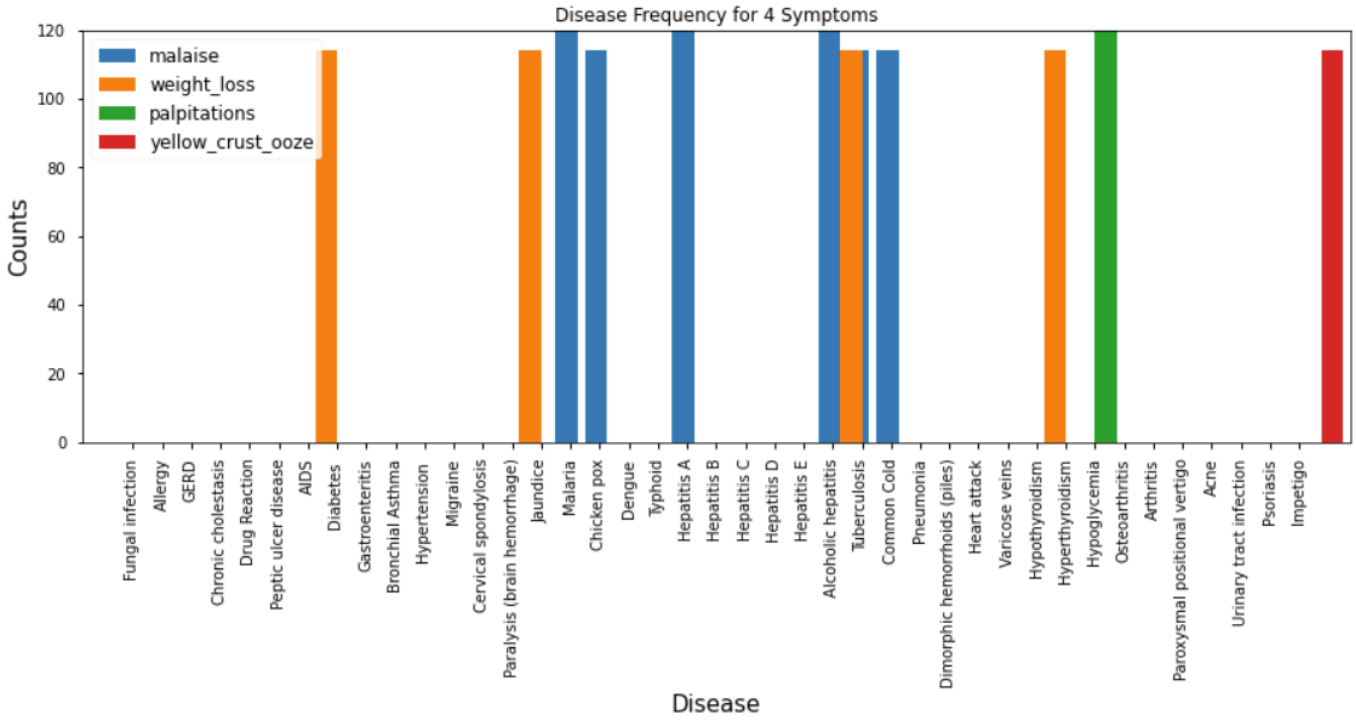

Figure 2: Frequency plot for the occurrence of each disease for symptoms 'malaise', 'weight_loss', 'palpitations', and 'yellow_crust_ooze'. These 4 symptoms are representative of the 4 categories described in the section above.

Predictive Power Score of Each Symptom:

Calculating the Predictive Power Score of each feature on the target variable was conducted using the ppscore API available in Python from 8080 Labs[1]. This measure was an attractive option to quantify the correlation of each feature on the categorical target variable. A ppscore of 1 indicates that a feature column has perfect predictive power, while a score of 0 indicates a feature column has no predictive power. The maximum predictive power score was found to be 0.0041, and 20 symptoms scored here. 51 of the symptoms had a predictive power score of 0.00, indicating no predictive power. The remaining 60 symptoms scored between 0.0035 and 0.0021. This test showed us that no one feature was strongly correlated with the target feature ('prognosis'), and, indeed, that no feature on its own has strong predictive power. If dimensionality reduction was desired, the 51 features with ppscore of 0 would be good candidates, however in the machine learning models we tested there was no need for this. These results are summarized in Table 4, below.

Table 4: Summary of Predictive Power Scores for Each Symptom Feature on Target Variable

Predictive Power Score	0.0041	0.0035 – 0.0021	0
Number of Symptom Features	20	60	51
Interpretation	Best predictive power	Some predictive power	No predictive power

[1] 8080 Labs PPS Repo can be found on Github at this address: https://github.com/8080labs/ppscore

Data Preprocessing and Training Data Development

Data Encoding:

The symptom feature columns contained binary data, which were already formatted similarly to one-hot encoding. The target feature column contained categorical data, and was label encoded to map each distinct disease category to a distinct value for use with machine learning modeling.

Data Splitting:

The data were split into X and y, with X holding the 131 binary symptom features and y holding the label encoded categorical target feature. The data were further split into a training and testing set, with 80% of the data retained for training the machine learning models and 20% being retained to test each model's performance.

Modeling

Machine Learning Algorithms Implemented in SciKitLearn:

A total of 7 machine learning models were fit to the training data and tested on the test data: An entropy-based decision tree, a gini-impurity based decision tree, a random forest classifier, an XGBoost classifier, a gradient boosted classifier, and Ada boosted classifier and a support vector classifier with an RBF kernel. In each case, an "off-the-shelf" version of the model was tried first before attempting hyperparameter tuning. The exceptions to this rule were the entropy-based and gini-impurity based decision tree models, which were tuned in their first implementation due to commonly known overfitting issues in unbound implementations. These models were compared on the basis of accuracy, F1 score, precision and recall on the test set. Ultimately, hyperparameter tuning was only performed on the decision tree classifiers and the Ada boost classifier, because every other classifier tested showed 100% accuracy on the test set. The test results are summarized in Table 5, below.

Table 5: Test Statistics for Tested Machine Learning Models

	Accuracy	F1	Precision	Recall
Random Forest	1.0	1.0	1.0	1.0
SVC	1.0	1.0	1.0	1.0
XGBoost	1.0	1.0	1.0	1.0
Gradient Boost	1.0	1.0	1.0	1.0
AdaBoost	0.99	0.99	0.99	0.99
Entropy Tree	0.95	0.95	0.96	0.96
Gini Tree	0.95	0.95	0.96	0.95

Model Selection:

Four models gave perfect accuracy, F1, precision, and recall on the test set: random forest, SVC, XGBoost, and Gradient Boost. While this indicated great success in the classification problem, it gave rise to new questions. Namely, did we make a mistake and accidentally leave the target feature column in the independent variable set? Two pieces of evidence suggested this was not the case:

1. The three lowest-performing models were below 100% accuracy. If the high accuracy of the best-performing models was an artifact of leaving the target variable in the independent variable feature space, we would expect all models to show 100% accuracy.
2. We investigated the feature importance values for one of the top performing models, the random forest classifier. The feature importance is a calculated value that explains how important a given feature is to the overall prediction; we expect a reasonable model to contain multiple feature importances, each a fraction of 1.0, the sum of which is 1.0. All feature importances of the random forest classifier were quite low and the sum across the feature space was 1.0, indicating that all features carried some weight in the classification and no one feature dominated the model's prediction. A histogram of the feature importances can be seen in Figure 3, below.

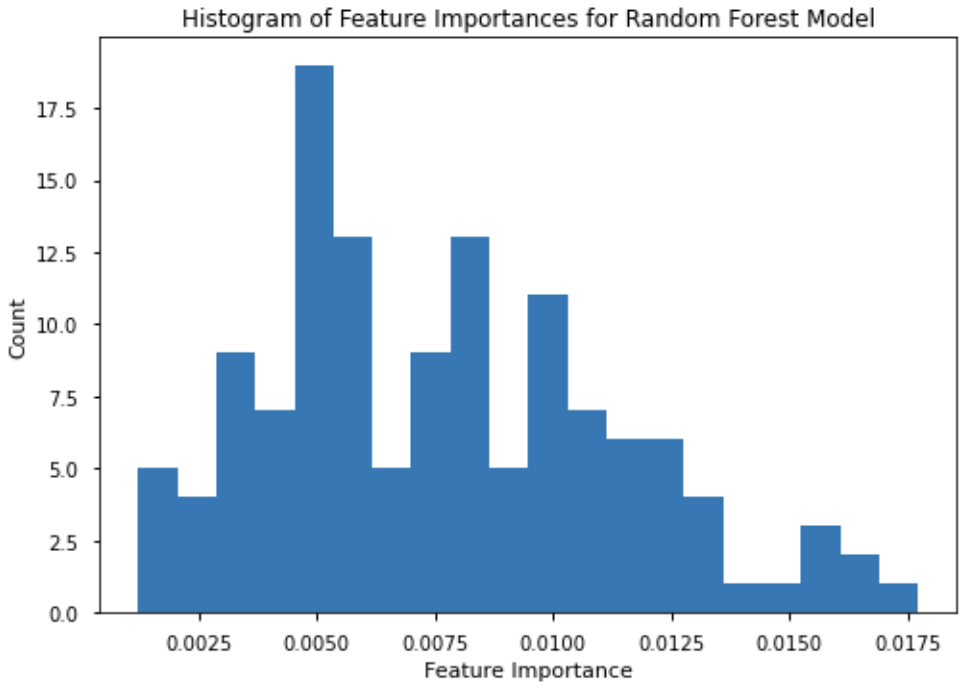

Figure 3: The histogram of feature importances shows that no one feature carried undue weight in the prediction of the classifier.

In order to make a selection on the "best model" out of the four top-performing models, we turned to another measure: model training time and sample prediction time. We measured both, with results summarized in Table 6, below.

If we deploy a model that continuously retrains every time new data comes in, then the training time metric would be the most important to optimize. Because this is a medical diagnostic tool, we would probably retrain in careful batches to maintain control over the model, so training time is less important. By this measure, the SVC Classifier is superior, taking just 0.59s to fit to our training data, and the Random Forest model is right behind at 0.61s.

We were more interested in how quickly a model could predict a diagnosis for a given occurrence. This metric would indicate how quickly a medical profession could expect to retrieve a diagnosis classification after inputting a patient's symptom data in the model in a production environment. By this measure, the XGBoost classifier shows superiority, with the lowest test time of just 0.02ms per patient.

The model chosen would depend on how the model is deployed: the XGBoost Classifier has the quickest prediction time, but 37x the training time compared to the SVC model. The training time might be prohibitively slow for retraining the model as the quantity of data increases, as it would in a production environment. This could potentially be offset by utilizing XGBoost's parallel processing feature in a production

environment, however that would take additional resources, and given equally accurate choices, it is good practice to choose that which can be implemented most simply.

Sitting in between XGBoost and SVC is the Random Forest Classifier. With a training time of 0.67s and test time of 0.05ms per patient, this model blends quick turn-around with quick training and is therefore the recommended model to move into production.

Table 6: Model Training Time and Sample Prediction Time Results		
	Training Time (s)	Prediction Time per Patient (ms)
Random Forest	0.61	0.04
SVC	0.59	0.09
XGBoost	21.59	0.02
Gradient Boost	38.56	0.06

Conclusions and Next Steps

The off-the-shelf machine learning models in sklearn by and large did amazingly well in predicting patient diagnoses in our testing data. We found 4 machine learning models that performed diagnosis classification at 100% accuracy on the test set, and selected the Random Forest Classifier as the recommended model to use in a production setting.

The 100% accuracy score is too good to be true in most settings, and certainly great caution must be taken when using Machine Learning to affect patient care. Therefore, this modeling process should be taken as **proof of concept** that such a diagnostic tool *can* be built to make accurate diagnoses, but further revision and testing with patient data is definitely advised. To improve the strength of the model, we suggest augmenting the dataset with more observations. As more data is gathered, the model can be retrained with the extensive real-world data to challenge the model and extend its functionality.

When pushing the model through to production, make sure to use LabelEncoder's inverse_transform function on the predicted values to return the interpretable disease names rather than label encoded values.

Extending this type of modeling into a recommender system or using predict_proba in sklearn to return a series of possible diagnoses with associated probabilities could help the diagnostic tool to be more readily accepted by practicing medical professionals. After all, diagnosis has been an art form, and diagnosticians are unlikely to happily give that up. However, a useful tool that could return several likely diagnoses in order of probability would still give the diagnostician some space for human interpretation. This would be good at least in the beginning of using such a tool, until widespread trust and proven effectiveness can be established with real patient data.