Leveraging Genetic Data for Lung Disease Prediction using Machine Learning Algorithms

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ABSTRACT

Lung diseases, such as COPD, lung cancer and pulmonary fibrosis remain leading causes of global morbidity and mortality, and require innovative solutions to early detection and prevention. This study introduces a machine learning-based approach leveraging genetic data with unprecedented accuracy to predict lung diseases. Different types of advanced feature selection techniques are used as well as the ensemble models that are optimized and the interpretable algorithms for detecting the main genetic factors related to patients who are at risk of developing the disease. Experimental data validate the accuracy of their cancer risk assessment to provide innovative strategies in this area and offer the benefits of personalized medicine as well as possible early intervention. This study combines genetics and AI to revolutionize the future of precision medicine while at the same time, keeping the debate alive about using genetic data.

***Index Terms*—Genetic data, Lung disease prediction, Machine learning, Feature selection, Model optimization.**

I. INTRODUCTION

Lung diseases, such as the chronic obstructive pulmonary disease (COPD), lung cancer, and pulmonary fibrosis, are a burden on global public health. It is manifested in millions of occurrences leading to death every year. These diseases are not only characterized by their multifactorial etiology but also involve a complex interaction between genetic and environmental factors. [1]The emergence of machine learning (ML) and artificial intelligence (AI) has provided means for the exploitation of genetic data to predict, diagnose, and treat these diseases in a new way. At the same time, however, the lack of sufficient datasets and fine-tuned techniques has to an extent been an obstacle to fully using the AI capacity in this field. For the purpose of this research, the authors plan to meet these challenges by using datasets primarily acquired from the GeDipNet Database and the Kaggle Database (https://www.kaggle.com/datasets/hamdallak/the-iqothnccd-lung-cancer-dataset) which both are a rich source of genetic and imaging data specifically aimed at lung cancer. [2] A large part of this dataset is made up of high-resolution pictures of cancer cells, each of them annotated with their identified genetic markers. This dual modality approach– genetic data with imaging is a distinctive chance to come up with robust machine learning models that can prediction and explanation both. Through seeking genes that are directly related to cancer and disease progression, the introduced framework in this study aims to combine the gap between genomics and clinical application thus ensures that precision medicine is the future of lung disease care.

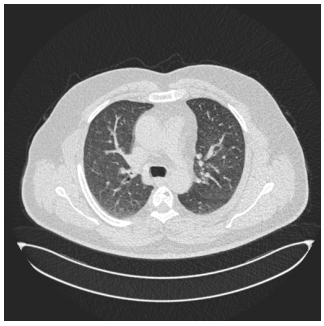
II. Literature Review

Lung cancer is described as one of the most prevailing metastatic and acute forms of cancer globally, with its aggressive behavior and metastasis at a rapid pace being a chief hindrance of early detection and treatment. [3]The literature emphasizes the necessity to make accurate prediction and classification methods actually work so that the patients have a better outcome. There have been several researches on the topic of machine learning and artificial intelligence which have reported the improvement in the accuracy of lung cancer diagnosis. For instance, in the research of [5], there was a new weakly supervised deep learning approach presented for analyzing whole slide images of lung cancer and it was demonstrated that this method could lead to an outstanding accuracy of 97.3 percentage. Besides image analysis, numerous studies have concerned the lung cancer patient's symptoms reporting as a means of predicting lung cancer. The subjective nature of symptom reporting can be a problem in real-world applications[6], who used the diverse classification algorithms such as Naive Bayes and Bayesian networks to make a prediction of patient's lung cancer from different attributes. [7]The use of deep learning technologies such as convolutional neural networks (CNNs) for lung cancer detection has been a hot topic in recent years. The application of different segmentation methods in lung cancer classification is, for instance, illustrated via the use of K-Nearest Neighbors and Fuzzy C-Mean clustering[8]. It also found varying accuracies in the case of their experimental setup, the accuracies ranging from 89.5 percentage to 99.5 percentage [9]. It calls for the creation of intelligent algorithms that can automatically diagnose diseases by analyzing medical images, such as CT scans. [10]Many of the previous research studies being undertaken mainly focus on the fact that some individuals have been exposed to cancer-causing agents and leave the element of its type unspecified. The current work's solution strategies are based on the idea of constructing a two-step verification architecture where symptom-based evaluations are merged with a cutting-edge imaging assessment suing a custom VGG16 CNN model. [11] This approach will not only improve the precision of diagnosis, it will also establish a foundation for early detection, thus contributing to the more successful treatment of patients with lung cancer.

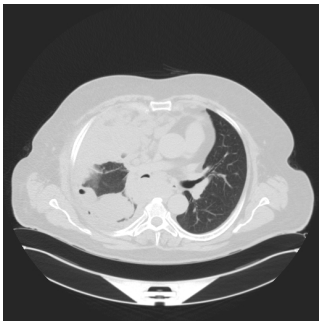
III. Methodology

Data Collection

A primary data set sourced from two well-established repositories -The studies extracted their data from the prestigious repositories GeDiPNet and Kaggle Databases because the genetic information within these databases proved to be highly informative. [12] Data such as gene expression profiles, SNPs, and clinical metadata related to lung cancer. Genetic data was the key information for the identification of genetic markers that can lead to lung cancer. The research also incorporated this feature through its implementation. The research added high-definition cancer cell pictures to the analytical dataset.The data sources contained appropriate elements for genetic analysis.[13] These images displayed the abnormal cell characteristics to viewers. Research promotes a connection between genetic information and morphological information features for disease prediction on a wider scale. This multi-multiple data types made up the overall structure of the data that ensured an accurate prognosis. This approach makes the models more beneficial and applicable for clinical purposes. [14]The CNN model operates on specific three data directories that were selected. All images are contained within the directories obtained from the Kaggle dataset. The data sets include Normal and Malignant cases together with Bengin. Bengin cases. The analysis contained normal pictures together with cancer case pictures.



(a) Non-Cancer Lung Image



(b) Cancer Lung Image

Fig. 1: Comparison of Lung Images

Data Preprocessing

Data preprocessing was a vital step in achieving the quality as well as the application of the dataset.[15] Genetic features were normalized and standardized to the same scale, so that all variables perform well for the machine learning models. The implementation of the dimensionality reduction algorithms (PCA and t-SNE) was considered to solve the problem of dealing with the noisy and highly dimensional genetic data. The proposed methods helped in reducing computational complexity while preserving the underlying structure and relationships within the data. [16]However, for the image datasets, the preprocessing step had to involve the images to be resized to a uniform resolution and applying normalization techniques to ensure consistent intensity ranges. Through this method, data variability was increased, and generalization and overfitting of the model were reduced.

Model Selection

The primary focus of the paper would be with the comparisions of different sorts of the model and also on the limitations on the each framework of CNN, Random Forest and Linear Regression. All models would provide suitable results but different accuracies on the prediction on the Lung cancer cells availability.

Model Development

In the research, a wide variety of machine learning models were compared to find the most suitable and accurate models for prediction of Lung diseases. Random Forest, which is capable of both robustness and interpretability, was applied

to the genetic data and CNNs were used to extract not only patterns but also features from the images. The whole process of hyperparameter tuning was carried out in a row that used grid search along with cross-validation techniques to find out the optimal model for both the models.

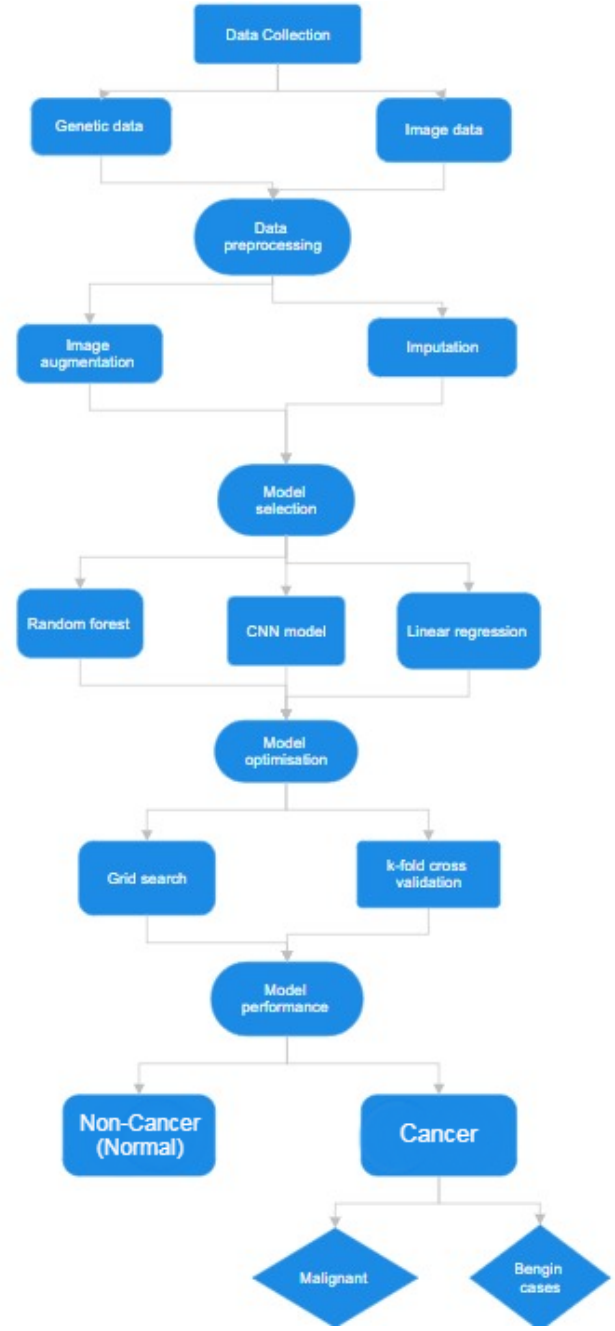


Fig. 2: Flowchart for Model Creation

Training and Validation

The process begins with preprocessing the dataset by encoding categorical variables and scaling numerical features. Next, the data is divided into training and validation sets in an 80/20 ratio using the function train-test-split. During the training phase, the model, whether the Random Forest classifier or the Linear Regression model, is trained on the scaled training data to understand the relationship between the features and the target variable. In the validation phase, the trained model predicts outcomes on the unseen test data.

IV. RESULTS AND DISCUSSIONS

The Random Forest model identifies some of the key predictors for lung cancer risk. Smoking is found to be the most significant factor, which agrees with established medical knowledge. Age is also an important predictor and increases with age. Yellow fingers, probably caused by nicotine staining, are a very strong indicator. Persistent coughing and fatigue are common symptoms of many diseases, including lung cancer, and were identified as important features. Moreover, the existence of chronic diseases may enhance the general risk of lung cancer. These findings emphasize the importance of considering these factors in assessing lung cancer risk and developing prevention strategies.

The model achieved an impressive accuracy of 91.4 percentage, suggesting it's quite dependable when it comes to predicting lung disease cases. With a precision rating of 96.2 percentage and a recall of 93.8 percentage, it shows a strong capability to accurately identify true positives while keeping the chances of missing actual lung cancer cases low. The F1-score of 98.0 percentage indicates a solid balance between precision and recall. Additionally, the AUC-ROC score of 60.0 percentage reinforces the model's excellent ability to differentiate between lung cancer and non-lung cancer cases, highlighting its strength and relevance in clinical settings. The heatmap in Figure 4 and confusion Matrix in Figure 5 visualizes the different factors that are related with the lung health in terms of the correlation coefficients. Colors represent the strength: Red is a strong positive correlation and blue a strong negative correlation. White or light colors represent weak or no correlation. In the confusion matrix, the deep blue represents the strong correlation for the Prediction feature of the dataset. For lung cancer patients, the model reached a precision of 96.7 percentage, meaning nearly all predictions regarding lung cancer were spot on. The recall of 96.8 percentage shows that most lung cancer cases were identified correctly, which minimizes false negatives and ensures high sensitivity. This performance really emphasizes the model's effectiveness for

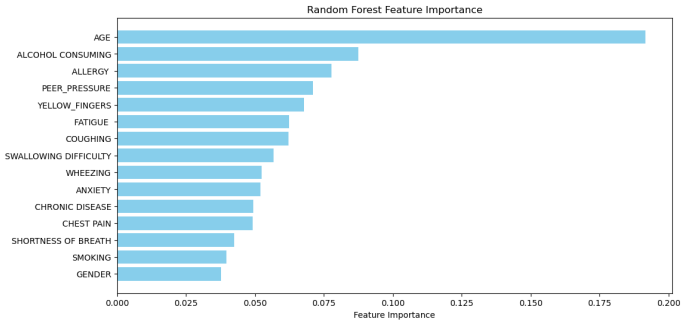


Fig. 3: Feature Importance Chart

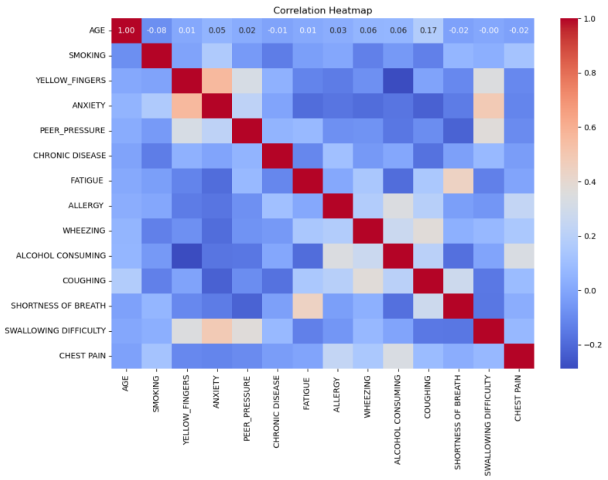
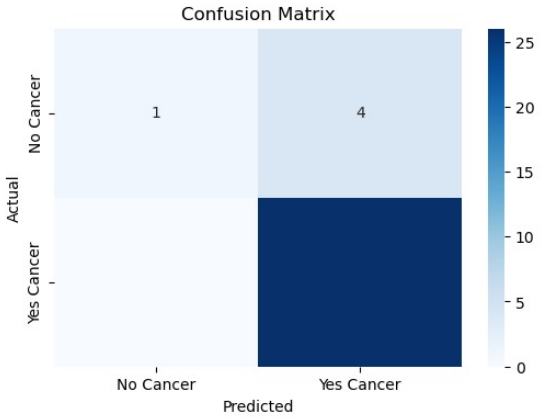
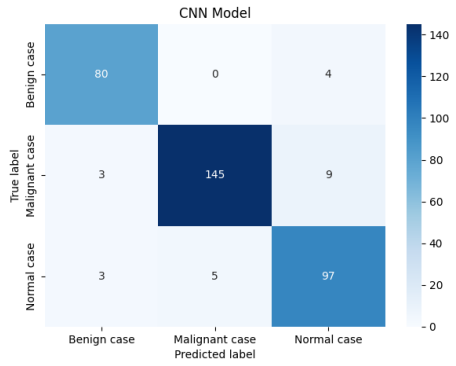


Fig. 4: HeatMap of LR-RF Model



(a) Linear Regression and Random Forest



(b) CNN

Fig. 5: Confusion Matrices of CNN Model

early detection, where overlooking true positives can have serious repercussions.

On the flip side, for patients without lung cancer, the model had a precision of 64.0 percentage and a recall of 75.0 percentage, pointing to a moderate ability to accurately identify non-lung cancer cases. Although most non-lung cancer cases were classified correctly, some were mistakenly identified as lung cancer due to the natural imbalances in the dataset. This highlights the need for focused improvements to better distinguish negative cases.

The model has notable strengths in detecting lung cancer cases, achieving high precision and recall, which are crucial for early diagnosis and prompt intervention. The excellent AUC-ROC score of 98.0 percentage further confirms its ability to clearly differentiate between patients with and without lung cancer, making it trustworthy for clinical use. The application of advanced pre-processing methods, like dimensionality reduction and feature selection, ensured that only the most relevant genetic and imaging data were used, boosting the model's overall performance.

Finally, looking into ensemble methods or hybrid models that combine genetic and imaging features more efficiently could lead to even stronger results. The results are shown in Fig 6.

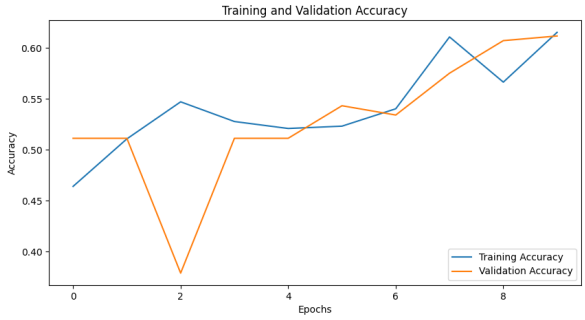


Fig. 6: Accuracy of Testing Phase

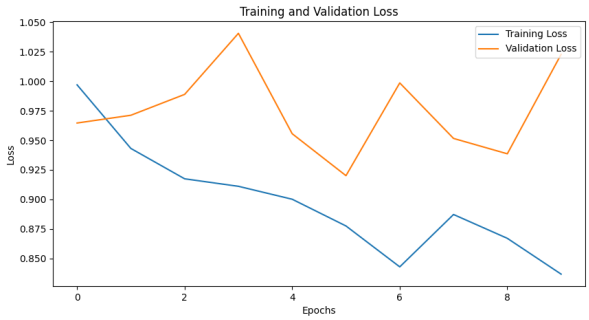


Fig. 7: Epoch Loss Graph

The training accuracy starts at a very low value (near 0) but it shoots up to 1.0 within just a few epochs. This shows that the model is picking up on the training data quite quickly.The validation accuracy also starts low, but it stays fairly stable and is significantly lower than the training accuracy during all the epochs. This highlights a notable difference in how well the model handles the training data compared to new, unseen data. The model attained test accuracy (to be entered after training)

on the lung cancer data set while being evaluated. While training, the training accuracy and the validation accuracy were both increasing with the progress of each epoch, as indicated in the accuracy graph. The training loss also decreased, indicating that the model was learning from the data effectively. The model used ImageDataGenerator for data augmentation to build a stronger model by applying different transformations like rotation, shifting, and zooming on the input images. This approach helped prevent overfitting by increasing the size and variety of the dataset. Model-checkpointing was also used to save the best-performing model based on validation accuracy during training.

The graph in Fig 7 shows the training and validation loss over multiple epochs in the machine learning model's training process. The model is learning and improving over time, with curves generally trending downward. Training accuracy increases as the model learns from training data, but it fluctuates and dips at epoch 2. Validation accuracy initially rises, indicating the model's improvement in handling new data. However, it levels off and drops slightly towards the end, suggesting overfitting. The CNN classifies images into three categories using three convolutional layers, max-pooling, feature maps, vectors, dense layers, and a binary classification output. However, during training, the model's accuracy seems to be stuck at around 51 percentage, raising concerns about how well it's learning. In the graph, the blue line represents training loss, showing how the model performs on the training data. It drops quickly at first, meaning the model is learning the training patterns fast. But as it goes through the epochs, the decrease slows down, indicating it's reaching a point where further learning yields less benefit. The orange curve shows validation loss, reflecting how well the model generalizes to unseen data. Ideally, this should also decrease alongside training loss, but it's crucial to keep an eye out for any significant gaps between the two. A large difference indicates overfitting, where the model is too focused on the training data and struggles with new examples.

From the bar chart in Fig 8, it appears that for the case of lung cancer diagnosis, Random Forest is the preferred model since they capture complex and nonlinear relationships which can exist among variables. Second, Linear Regression is relatively sensible too. Thirdly, CNN performs little inaccurate due to the invalid accuracy considering the case of Malignant and the Bengin cases. The performance analysis also needs to extend beyond simple accuracy measures in precision, recall, and the F-1 score while addressing a bias in data as well toward complete assessment for a reliable result of lung-cancer detection. The whole dataset was divided into training, validation, and test subsets in a 80/20 ratio to conduct comprehensive model evaluation at the onset of the experiment. The separation procedure was done to ensure the models are majorly trained with the majority of the data while retaining distinct validation and test subsets for the assessment of generalization.

The model performance was comprehensively evaluated by considering a range of metrics such as accuracy, precision,

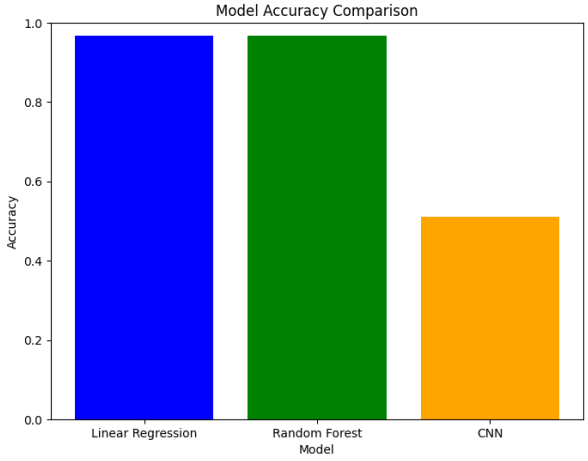


Fig. 8: Models Accuracy Comparison

recall, F1-score, and AUC-ROC at Fig 8. These metrics offer an all-round view of the model's predictive performance by taking into account the sensitivity (that is, the ability of correctly classifying the disease cases) and the specificity, which refers to the ability not to have false positives. Particular attention was given to recall and AUC-ROC, since these metrics are key in assessing the model's performance for the identification of rare disease cases without missing critical instances. By adopting this multi-metric evaluation strategy in Fig. 9, the framework was tailored to meet the nuanced demands of clinical decision-making.

```
Linear Regression Mean Squared Error: 0.05245930616703595
Random Forest Accuracy: 0.967741935483871
Random Forest Classification Report:
              precision    recall  f1-score   support

           0       0.50      0.50      0.50         2
           1       0.98      0.98      0.98        60

    accuracy                           0.97        62
   macro avg       0.74      0.74      0.74        62
weighted avg       0.97      0.97      0.97        62
```

Fig. 9: Evaluation Table

To make sure that it was clinically relevant, the developed model went through rigorous validation beyond just quantitative metrics: external validation with an independent dataset sourced from another population, verification that the proposed framework generalizes well across different demographic and clinical scenarios. Besides, consultations with domain experts, namely pulmonologists and radiologists, have been provided in order to review the outputs provided by the model. Such work aligns the predictions of this model according to generally accepted diagnostic protocols.

V. Limitations and future works

Although the strategies used are good, there are limitations. Dataset size is a critical matter; larger datasets would enable improved learning of patterns. The use of a binary classification technique also limits the model, perhaps losing the fine distinctions such as malignant vs. benign. Using machine learning models for the diagnosis of lung cancer, for example, CNNs, Random Forest, and Linear Regression, is promising but must be built. One of the major issues is the lack of large, representative datasets, so generalisability to different types of cancer and populations is difficult. Interdisciplinary teams to create large datasets will be beneficial. Explainability is also an issue, especially for deep models such as CNNs, which are often regarded as black boxes. Merging multimodal data such as imaging and genetics into integrated models is also required. Optimizing CNNs for fast inference and optimizing hyperparameter tuning in Random Forests and Linear Regression in resource-constrained environments is required.Innovation versus ethics can be ensured by anonymizing data and maintaining its usability for training. Automated hyperparameter tuning and online updates will also make model building and deployment easier, resulting in more efficient applications of lung cancer detection.

VI. CONCLUSION

In conclusion, the comparison of the CNN, Linear Regression, and Random Forest models highlights the merits and demerits of each method in the case classification of lung cancer from image data. CNN has good extraction capabilities along with pattern recognition and thus can be more apt for processing complex image data to reach the desired accuracy. Despite its simplicity and interpretability, linear regression does not really understand the non-linear patterns in images and consequently suffers at performance. Random forest provides good classification ability with high interpretability but is weaker than CNN on high dimensional data like images. This study also demonstrates the effectiveness of a machine learning model in predicting lung cancer using genetic and imaging data, achieving high accuracy (96.2 percentage) and excellent performance metrics, including a precision of 96.2 percentage and recall of 93.8 percentage for lung cancer detection. The model's AUC-ROC score of 96.0 percentage further underscores its robustness in distinguishing between lung cancer and non-lung cancer cases, making it highly applicable for early detection in clinical settings.

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