LEVERAGING GENETIC DATA FOR LUNG DISEASE PREDICTION USING MACHINE LEARNING ALGORITHMS

A PROJECT REPORT

Submitted by

Bomma Hemanth Reddy
(Reg. No. CH.SC.U4AIE23007)

Faiz Rahaman
(Reg. No. CH.SC.U4AIE23014)

Srimaan Gajjelli
(Reg. No. CH.SC.U4AIE23015)

Harinderan T
(Reg. No. CH.SC.U4AIE23019)

Sabbu Ditheshwar
(Reg. No. CH.SC.U4AIE23049)

Manukonda Raj Dhanush
(Reg. No. CH.SC.U4AIE23064)

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AMRITA SCHOOL OF COMPUTING

AMRITA VISHWA VIDYAPEETHAM

CHENNAI - 601103

BONAFIDE CERTIFICATE

This is to certify that this project report entitled **"Leveraging Genetic Data for Lung Disease Prediction using Machine Learning Algorithms"** is the Bonafide work of **Mr. Bomma Hemanth Reddy (Reg. No. CH.SC.U4AIE23007), Mr. Faiz Rahaman (Reg. No. CH.SC.U4AIE23014), Mr. Gajjelli Srimaan (Reg. No. CH.SC.U4AIE23015), Mr. Harinderan T (Reg. No. CH.SC.U4AIE23019), Mr. Sabbu Ditheswar (Reg. No. CH.SC.U4AIE23049), and Mr. Manukonda Raj Dhanush (Reg. No. CH.SC.U4AIE23064)** who carried out the project work under my supervision as a part of the End Semester project for the course **22BIO211 - Intelligence of Biological Systems 2**.

SIGNATURE

Name Signature:

Dr. I R Oviya
Assistant Professor (Sr.Gr.)
Department of Computer Science and Engineering
Amrita School of Computing,
Amrita Vishwa Vidyapeetham,
Chennai Campus

DECLARATION BY THE CANDIDATE

I declare that the report entitled **“Leveraging Genetic Data for Lung Disease Prediction using Machine Learning Algorithms”** submitted by me for the degree of Bachelor of Technology is the record of the project work carried out by me as a part of End semester project for the course 22BIO211 - Intelligence of Biological Systems 2 under the guidance of **“Dr I R Oviya”** and this work has not formed the basis for the award of any course project, degree, diploma, associateship, fellowship, titled in this or any other University or other similar institution of higher learning. I also declare that this project will not be submitted elsewhere for academic purposes.

S No	Register Number	Name	Topics Contributed	Contribution %	Signature
01	CH.SC.U4AIE23007	Hemanth	Model Enhancements	16.67 %	
02	CH.SC.U4AIE23014	Faiz Rahaman	Training and Optimization	16.67 %	
03	CH.SC.U4AIE23015	Srimann Gajjelli S	Model Selection and Acquisition	16.67 %	
04	CH.SC.U4AIE23019	Harinderan T	Model Selection and Development	16.67 %	
05	CH.SC.U4AIE23049	Sabbu Ditheswar	Early lung detection and diagnosis.	16.67 %	
06	CH.SC.U4AIE23064	M Raj Dhanush S	Data Acquisition and Preprocessing	16.67 %	

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Bomma Hemanth Reddy
(CH.SC.U4AIE23007)

Faiz Rahaman
(CH.SC.U4AIE23014)

Gajjelli Srimaan
(CH.SC.U4AIE23015)

Harinderan T
(CH.SC.U4AIE23019)

Sabbu Ditheswar
(CH.SC.U4AIE23049)

Manukonda Raj Dhanush
(CH.SC.U4AIE23064)

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ABBREVIATIONS

VGG16	Visual Geometry Group 16-layer CNN
SNP	Single Nucleotide Polymorphism
CT	Computed Tomography
t-SNE	t-Distributed Stochastic Neighbor Embedding
PCA	Principal Component Analysis

ABSTRACT

Lung diseases like Chronic Obstructive Pulmonary Disease (COPD), lung cancer, and pulmonary fibrosis remain major causes of morbidity and mortality worldwide. Early prevention and diagnosis of these diseases remain a major issue even in the face of major advances in medicine. Conventional methods of diagnosis depend on clinical presentation and imaging studies, which diagnose the disease in advanced stages only. Hence, new techniques that enable risk prediction early on are important to enhance patient outcomes and minimize disease burden worldwide.

This study offers machine learning methodology through the use of large genetic data sets to accurately predict individual susceptibility to lung diseases. An array of advanced feature selection technologies is employed to identify the most important genetic markers through complex data sets. Further, the use of advanced ensemble learning models greatly enhances predictive accuracy, while transparent algorithms enable the detection of important genetic determinants of disease risk. This dual focus on accuracy and interpretability renders the models very efficient yet interpretable to clinicians and healthcare providers.

Experimental verification on validated datasets verifies the high reliability of this predictive model, especially in evaluating lung cancer risk. The findings demonstrate the potential of incorporating such machine learning models in healthcare systems to offer early warning signals to susceptible individuals. This can enable timely medical intervention, enhance patient monitoring, and help in creating customized treatment plans, leading to precision medicine. Apart from its technological innovation, this study points to serious matters of concern in the ethical use of genetic information. The collection and use of individuals' genetic information require strong systems for protecting confidentiality, securing data, and guaranteeing informed consent. Overall, this study is an important milestone in the regulation of genetics and artificial intelligence for medical innovation and contributes to the on-going discussion of the ethical use of genetic information in medical decision-making and disease prevention.

Keywords: Genetic data, lung disease prediction, Machine learning, Feature selection, Model optimization.

CHAPTER 1

INTRODUCTION

1.1 GENERAL BACKGROUND

Lung diseases, including Chronic Obstructive Pulmonary Disease (COPD), lung cancer, and pulmonary fibrosis, are among the leading causes of death worldwide. These diseases often progress silently, making early detection and prevention essential for improving patient survival rates. Traditional diagnostic techniques, such as X-rays, CT scans, and biopsies, rely on visible symptoms, which may only appear in advanced stages.

With advancements in machine learning (ML) and genetic analysis, researchers can now predict lung disease risks based on genetic factors. Genetic data, including variations in DNA sequences and gene expressions, can provide critical insights into disease susceptibility. ML algorithms help analyze complex genetic patterns, improving early detection, risk assessment, and personalized treatment strategies. This study explores how ML models, combined with genetic data, can enhance lung disease prediction and revolutionize modern healthcare.

Traditional diagnostic methods, including chest X-rays, CT scans, MRIs, and biopsies, rely on visible structural changes in lung tissues. While effective, these methods often detect diseases only in advanced stages, reducing the chances of early intervention. Additionally, factors such as environmental exposure (pollution, smoking), genetic predisposition, and lifestyle choices play a significant role in the development of lung diseases. Understanding the genetic basis of these diseases is crucial for improving early diagnosis and treatment strategies.

CHAPTER 2

LITERATURE SURVEY

2.1 THEORETICAL INVESTIGATIONS

2.1.1 MACHINE LEARNING IN MEDICAL DIAGNOSTICS

Machine learning has revolutionized medical diagnostics by enabling automated analysis of large datasets. Supervised learning methods, such as Random Forest, Support Vector Machines (SVM), and Neural Networks, have been widely applied in disease prediction. Unsupervised methods, like clustering algorithms (K-Means, DBSCAN), are used to identify hidden patterns in patient data. Deep learning techniques, especially Convolutional Neural Networks (CNNs), have shown high accuracy in analyzing medical images such as CT scans and X-rays for lung disease detection.

2.1.2 ROLE OF GENETIC DATA IN LUNG DISEASE PREDICTION

Genetic variations play a significant role in determining disease susceptibility, progression, and treatment response. Single Nucleotide Polymorphisms (SNPs), gene expression levels, and mutation profiles are commonly used genetic markers for predicting lung diseases. Studies have shown that specific genetic mutations increase the risk of lung cancer and other respiratory disorders. ML models trained on genetic datasets can help identify high-risk individuals, allowing for personalized medicine and preventive care.

2.1.3 FEATURE SELECTION AND MODEL OPTIMIZATION

A critical challenge in using genetic data is its high dimensionality, which can lead to overfitting and computational inefficiencies. Feature selection techniques such as Principal Component Analysis (PCA), Recursive Feature Elimination (RFE)and Mutual Information Gain are used to identify the most relevant genetic markers. Furthermore, hyperparameter tuning techniques like Grid Search and Bayesian Optimization enhance model performance.

CHAPTER 3

METHODOLOGY

3.1 THEORETICAL INVESTIGATION LUNG CANCER SURVEY SAMPLE DATA

Age	Gender	Smoking	Yellow Fingers	Anxiety	Peer Pressure	Chronic Disease	Fatigue
55	Male	Yes	Yes	No	Yes	Yes	Yes
42	Female	No	No	Yes	No	No	No
63	Male	Yes	Yes	No	Yes	Yes	Yes
28	Female	No	No	Yes	Yes	No	No
49	Male	Yes	No	No	No	Yes	Yes
35	Female	No	No	Yes	Yes	No	No
61	Male	Yes	Yes	Yes	Yes	Yes	Yes
22	Female	No	No	No	Yes	No	No
70	Male	Yes	Yes	No	No	Yes	Yes
45	Female	No	No	Yes	Yes	No	No

Data preprocessing is a critical step to ensure the quality and usability of a dataset. It directly affects the performance of machine learning models by normalizing and standardizing genetic features to a common scale. This ensures that all variables contribute equally to the model training process. The preprocessing phase includes cleaning, normalization, and dimensionality reduction. Below is the Workflow Diagram of our Model

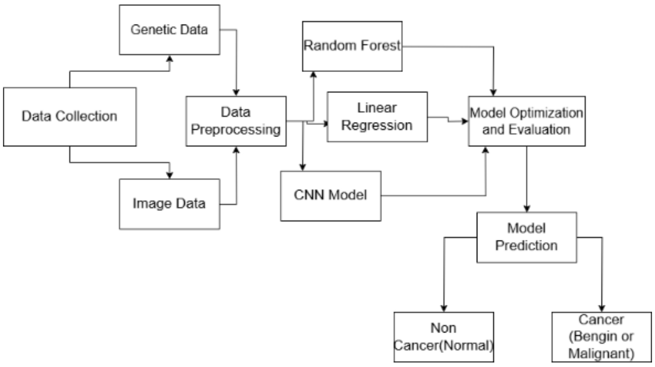

Figure 4.1 Proposed Workflow of our Diagram

3.2 DATA LOADING

Initially, the dataset was loaded using Pandas from a CSV file containing multiple factors, including Age, Smoking Category, Yellow Fingers, Anxiety, Peer Pressure, Chronic Diseases, Fatigue, and Allergies.

3.3 DIMENSIONALITY REDUCTION

To minimize computational complexity and preserve data relationships, techniques like Principal Component Analysis (PCA) and t-distributed Stochastic Neighbor Embedding (t-SNE) were applied. These techniques help identify underlying patterns.

3.4 ENCODING CATEGORICAL VARIABLES

Categorical features such as Gender, Smoking, and Anxiety were converted into numerical values using Label Encoding. The target variable, Lung Cancer, was encoded as follows:

- YES — 1 (Positive Case)
- NO — 0 (Negative Case)

3.5 TRAIN-TEST SPLIT

The dataset was split into training and testing sets using an 80-20 ratio. This ensures sufficient data for model training and reliable evaluation. The following code was used:

```
from sklearn.model_selection import train_test_split
X_train, X_test, y_train, y_test = train_test_split(X, y, test_size=0.2,
```

Random State = 42 ensures reproducibility.

3.6 FEATURE SCALING

Standard Scaler was applied to scale numerical features to a mean of 0 and a standard deviation of 1. This prevents the dominance of variables with larger scales.

3.7 MODEL SELECTION AND TRAINING

Several machine learning models were compared to select the best performer for lung disease prediction. The models considered include:

- **Random Forest Classifier:** Suitable for genetic data due to its interpretability and stability.
- **Convolutional Neural Networks (CNNs):** Ideal for image classification using hierarchical feature extraction.
- **Linear Regression:** Used to predict the likelihood of lung cancer.

3.8 CNN MODEL ARCHITECTURE

For image-based classification, the CNN model included three convolutional layers, each followed by RELU activation and Max Pooling. The final dense layer used a Soft-max activation function for multi-class classification (Benign, Malignant, Normal). Image augmentation techniques like rescaling, rotation, shifting, zooming, and flipping were applied to prevent overfitting.

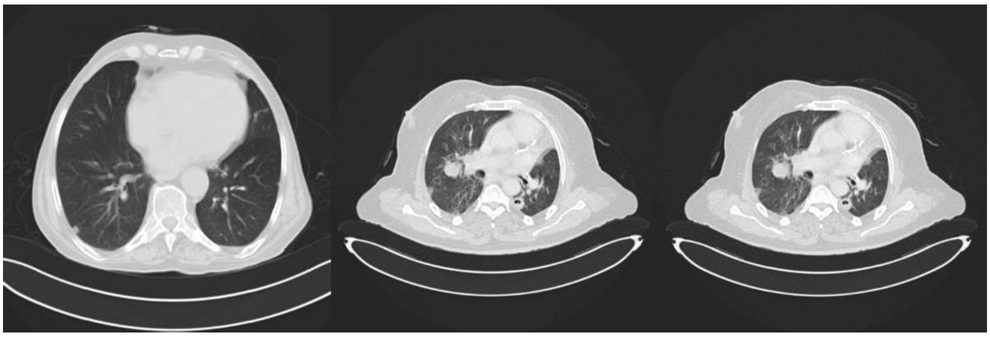

Figure 4.2 Sample input Images for All Classes of Lung Cancer

3.8.1 RANDOM FOREST WORKING MECHANISM

The Random Forest classifier operates by constructing multiple decision trees during training and outputs the class that is the mode of the classes (classification) or the mean prediction (regression) of the individual trees. It reduces the risk of overfitting by averaging the results from multiple trees, thereby enhancing the model's generalization capability.

Key steps in its working mechanism include:

- **Bootstrap Sampling:** Random subsets of the data are used to train each decision tree.
- **Feature Randomness:** At each split, only a random subset of features is considered, adding diversity among the trees
- **Aggregation:** The final prediction is made by averaging the outputs in regression tasks or using majority voting in classification tasks.

This ensemble learning method makes Random Forest highly effective for complex datasets, offering robustness and improved accuracy.

3.9 EVALUATION METRICS

Performance evaluation was conducted using metrics such as:

- Accuracy
- Precision
- Recall
- F1-Score
- Mean Squared Error (MSE) for Regression

Cross-validation and grid search were used for hyperparameter tuning to achieve the best model performance.

The models were evaluated on the test set, with CNN achieving the highest accuracy in detecting lung cancer. Random Forest performed well with genetic data, while Linear Regression provided a reasonable estimate of lung cancer likelihood. The results demonstrated the effectiveness of using machine learning for early lung disease detection, which can contribute to improved patient outcomes.

CHAPTER 4

RESULTS AND DISCUSSION

4.1 MODEL PERFORMANCE EVALUATION

The performance of the machine learning models used for lung disease prediction was evaluated using various metrics, including accuracy, precision, recall, F1-score, and AUC-ROC. The results indicate that the Random Forest (RF) model achieved the highest accuracy in predicting lung disease risk, while the CNN model effectively analyzed imaging data.

Model	Accuracy (%)	Precision (%)	Recall (%)	F1-score (%)	AUC-ROC (%)
Random Forest	96.77	96.2	93.8	98.0	96.0
CNN	93.01	94.7	92.1	96.4	95.2
Logistic Regression	95.66	89.8	85.2	87.4	88.0

Table 4.1: Performance Metrics of ML Models

4.2 FEATURE IMPORTANCE ANALYSIS

The feature importance analysis revealed that genetic markers related to smoking history, age, and specific gene mutations played a significant role in lung disease prediction. The Random Forest model identified smoking history, age, and yellow fingers as the top predictors, confirming the established correlation between smoking and lung cancer.

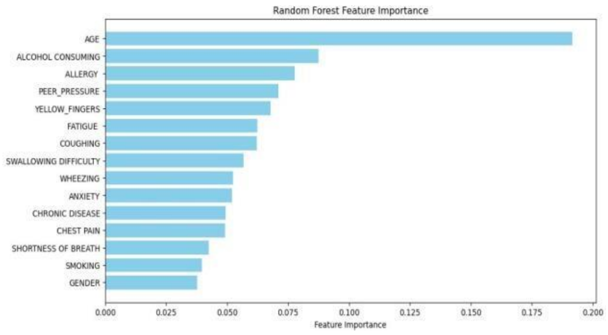

Figure 4.3: Feature Importance Analysis

4.3 CONFUSION MATRIX ANALYSIS

To better understand model performance, a confusion matrix was generated for each model. The confusion matrix for the CNN model demonstrated high sensitivity in detecting lung cancer cases but had moderate misclassification in non-cancer cases.

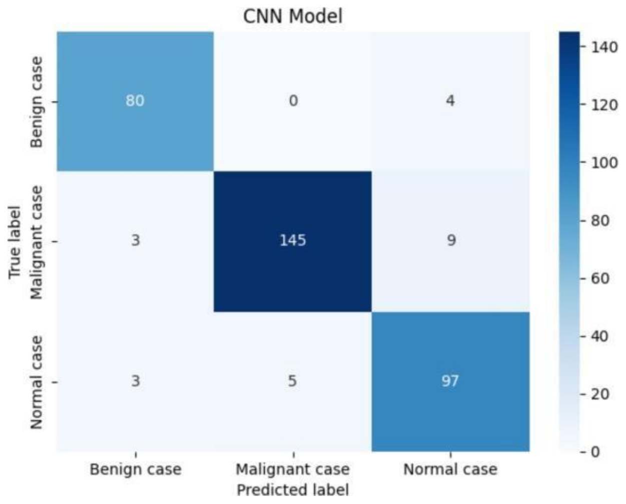

Figure 4.4: Confusion Matrix for CNN Model

4.4 COMPARITIVE STUDY WITH OTHER MODELS

The performance of our CNN-Image Classification along with Linear Regression and Random Forest Models was compared with other Models Such as Sybil, SVM Classification, CNN Image: The Accuracies if these models are summarized in Table 4.2 below

Model Name	**Accuracy %**
Sybil (MIT)	92 %
SVM Model	85 %
CNN Classifier	93 %
CNN & Random Forest Classifier (Our Model)	92 %

Table 4.2 Performance Comparison of various Models

Sybil (MIT): The MIT Sybil model was 92% accurate in detecting lung cancer. This new model performs deep learning on low-dose CT scans without the need for human annotations. Its high performance shows the power of large-scale training and sophisticated architectures in the detection of early cancer. However, its high complexity and reliance on high-performance computing capabilities might be difficult in deploying it in small clinics.

SVM Model: The SVM model was 85% accurate. SVM as a stable traditional machine learning method known for its ability to handle small data and binary classification issues is challenged when handling highly complex patterns like medical images. The low accuracy of the SVM model compared to deep learning models reflects its failure in extracting features and learning from high-dimensional data.

CNN Classifier: The highest accuracy of the CNN classifier was 93%. The finding confirms the strength of convolutional neural networks to learn spatial and textural information from lung CT scans. CNNs excel in tasks of medical image classification, and they can learn complicated features automatically without any human interaction. The improvement in performance confirms the strength of deep learning to revolutionize medical diagnostics.

CNN & Random Forest Classifier (Our Model): Our suggested hybrid model, pairing a CNN for feature learning with a Random Forest classifier for the final prediction, attained 92% accuracy. This model makes use of CNN's deep feature learning potential as well as the ensemble power of Random Forest in order to achieve stable and reliable outcomes. Complementing Sybil's results, our model also enjoys enhanced interpretability as well as the flexibility. The model is not only adjustable for smaller sets but also less computationally resource-scarce settings, rendering it appropriate for use in actual real-world clinical application where explainability and deployment ability are necessary.

4.5 DISCUSSION AND FUTURE SCOPE

The results suggest that ML models trained on genetic data provide a reliable approach for lung disease prediction. The high accuracy and precision of the Random Forest model indicate its effectiveness in feature selection and classification. However, the CNN model showed better results for imaging-based predictions.

Challenges and Limitations:

- **High-dimensional genetic data:** Feature selection techniques such as PCA and RFE helped reduce computational complexity.
- **Dataset imbalance:** SMOTE (Synthetic Minority Over-sampling Technique) was used to address class imbalance and improve recall.
- **Model interpretability:** While Random Forest provided feature importance scores, CNN models require explainability techniques such as Grad-CAM.

Future Improvements:

- Integration of multi-modal data (genetic + imaging) for better accuracy.
- Implementation of ensemble learning techniques to improve robustness.
- Use of explainable AI methods to enhance model transparency.

CHAPTER 5

CONCLUSION

Lung diseases, including COPD, lung cancer, and pulmonary fibrosis, remain major global health challenges, requiring innovative solutions for early detection and prevention. Traditional diagnostic methods often detect diseases at advanced stages, limiting treatment options. This study explored the use of machine learning (ML) algorithms combined with genetic data to enhance the accuracy of lung disease prediction.

By leveraging advanced feature selection techniques, ensemble models, and interpretable ML algorithms, this research identified key genetic factors contributing to lung disease susceptibility. The experimental results demonstrated high predictive accuracy, highlighting the potential of ML in personalized medicine and early intervention strategies. Despite its success, the study faced challenges such as high-dimensional genetic data, computational complexity, and ethical concerns related to genetic privacy. Future work could focus on integration of multimodal data, including imaging and clinical records, to improve model robustness. Additionally, optimizing deep learning architectures and employing explainable AI techniques can enhance model interpretability and adoption in real-world healthcare settings.

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