International Journal of Scientific Research in Engineering and Management (IJSREM)
Volume: 09 Issue: 04 | April - 2025 SJIF Rating: 8.586 ISSN: 2582-3930

Automatic Detection of Genetic Diseases in Pediatric Age Using Pupillometry

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***Abstract*— This paper introduces a comprehensive framework for automated disease detection using pupillometry data. Our approach establishes a robust pipeline that includes data preprocessing, feature extraction, and machine learning-based classification of patients based on their pupillary responses. We extract key features from both left and right pupil diameter measurements, such as maximum and minimum values, delta, channel height (CH), latency, and mean change velocity (MCV).To enhance classification accuracy, we train and evaluate multiple machine learning models, including Support Vector Machines (SVMs), an ensemble classifier, Extreme Learning Machines (ELM), Multi-Layer Perceptrons (MLPs), and Random Forests. Additionally, we propose a novel hybrid model that integrates the strengths of multiple algorithms, outperforming individual models in accuracy. Our experimental results highlight the effectiveness of this hybrid approach, demonstrating its potential for improving non-invasive and efficient disease diagnosis. This research contributes to advancements in clinical ophthalmology and neurology by leveraging pupillometry and machine learning for more precise and accessible diagnostic tools.**

Keywords: *Pupillometry data, Channel height, Latency, Mean Change Velocity, Support Vector Machines, Extreme Learning Machines, Multi Layer Perceptrons, Random Forest.*

I. INTRODUCTION

Pupillometry, the study of pupil size and reactivity, has gained attention as a promising non-invasive tool for assessing neurological and physiological functions. The pupil's response to various stimuli—such as light, cognitive tasks, and emotional cues—offers valuable insights into brain activity and the autonomic nervous system. Traditional diagnostic methods often rely on invasive procedures or subjective evaluations, emphasizing the need for objective and efficient alternatives. This research explores how pupillometry data, combined with advanced machine learning techniques, can automate disease detection and enhance diagnostic accuracy.[1]

Given the complexity and high dimensionality of pupillometry data, sophisticated analytical methods are required. Machine learning algorithms, known for their ability to identify intricate patterns and relationships, provide an effective approach for extracting meaningful information from such datasets. This paper presents a comprehensive pipeline that integrates data preprocessing, feature extraction, and model training to classify patients based on their pupillary responses[4]. By analyzing key features—including maximum and minimum pupil diameter,

delta, channel height, latency, and mean change velocity—our approach aims to capture critical pupillary dynamics associated with specific disease states[4].

This study evaluates and compares the performance of multiple machine learning models, such as Support Vector Machines (SVMs), ensemble classifiers, Extreme Learning Machines (ELM), Multi-Layer Perceptrons (MLPs), Random Forests, and a novel hybrid model, in the context of disease detection using pupillometry data. The goal is to determine the most effective modeling approach and establish a reliable framework for automated diagnosis. The findings of this research have the potential to transform clinical practice by enabling earlier and more precise disease detection, ultimately leading to better patient outcomes[8].

II. LITERATURE SURVEY

A study conducted by Ernesto Iadanza (Senior Member, IEEE)[1], Francesco Goretti, Michele Sorelli, Paolo Melillo (Member, IEEE)[2], Leandro Pecchia (Member, IEEE), Francesca Simonelli, and Monica Gherardelli examined pupillary responses to chromatic stimulation in patients with Retinitis Pigmentosa (RP) and healthy individuals.[6] The researchers employed a standardized protocol using a customized DP-2000 binocular pupillometer, which allowed for precise measurements of pupil dynamics in response to red, green, blue, and white light stimuli. Their meticulous experimental setup included a 10-minute dark adaptation period followed by a structured sequence of light stimulations. With a 30-Hz frame rate and high spatial resolution, the study captured detailed pupillary response traces while addressing data quality challenges by eliminating corrupted signals caused by blinking or eye movements. This rigorous methodology, emphasizing chromatic stimulation and high-fidelity data acquisition, lays a strong foundation for understanding disease-specific pupillary responses.[9]

Building upon the standardized protocols and meticulous data acquisition of the RP study, subsequent research has expanded the applications of pupillometry beyond ocular diseases[3]. While the RP study focused on chromatic light stimulation for a specific condition, other investigations have leveraged pupillometry to detect a wider range of neurological and systemic disorders. Researchers have applied various machine learning techniques, including Support Vector Machines (SVMs), neural networks, and ensemble methods, to analyze pupillary responses and classify disease states. Many of these studies utilize feature extraction techniques similar to those in the RP study—such as latency, contraction velocity, and redilation

time—but also incorporate advanced analytical approaches like frequency domain analysis and wavelet transforms. The RP study's emphasis on data quality control, particularly in identifying and excluding corrupted signals, has influenced modern research by establishing robust preprocessing pipelines as a standard practice in pupillometry-based machine learning applications[2].

An exciting development in the field is the integration of pupillometry with other physiological data, such as heart rate variability and electroencephalography (EEG), to gain deeper insights into disease mechanisms[9]. This multi-modal approach enhances diagnostic precision by providing a more comprehensive understanding of underlying physiological processes. Additionally, the emergence of deep learning techniques, such as Convolutional Neural Networks (CNNs) and Recurrent Neural Networks (RNNs), has revolutionized pupillometry research by enabling automatic feature extraction from raw pupillary response data, reducing dependence on manual feature engineering[10]. While these advancements diverge from traditional SVM-based methods used in earlier studies, they reflect the evolving landscape of pupillometry research. The continued focus on standardized protocols, data quality, and advanced analytical techniques—pioneered by studies like the RP research—remains instrumental in shaping the future of pupillometry for disease detection and diagnosis[9].

III. EXISTING SYSTEM

The current approach to diagnosing genetic diseases in pediatric patients, particularly **Retinitis Pigmentosa (RP)**, relies heavily on traditional clinical evaluation methods. These diagnostic processes involve a complex combination of clinical tests, many of which can be invasive and challenging for children. Additionally, inherited retinal diseases (IRDs) are caused by a wide range of genetic mutations, making it difficult to conduct timely and comprehensive screening using conventional methods. As a result, early diagnosis and effective monitoring remain significant challenges in pediatric populations[5].

DISADVANTAGES OF EXISTING SYSTEM

Despite its widespread use, the current diagnostic system presents several limitations:

- **Invasive procedures**: Some clinical tests require invasive techniques, which may not be appropriate for infants or young children, causing discomfort and potential risks.
- **Time-consuming and expensive**: The combination of clinical and genetic testing is both **labor-intensive and costly**, making access to timely diagnosis difficult, especially in resource-limited settings.
- **Lack of conclusive results**: Even with advanced testing, **diagnostic uncertainty remains a challenge**. In many cases, patients require additional testing, leading to delays in treatment and increasing the burden on healthcare systems.

Given these limitations, there is a growing need for **non-invasive, efficient, and cost-effective diagnostic alternatives** that can improve early detection and monitoring of genetic diseases in pediatric patients.

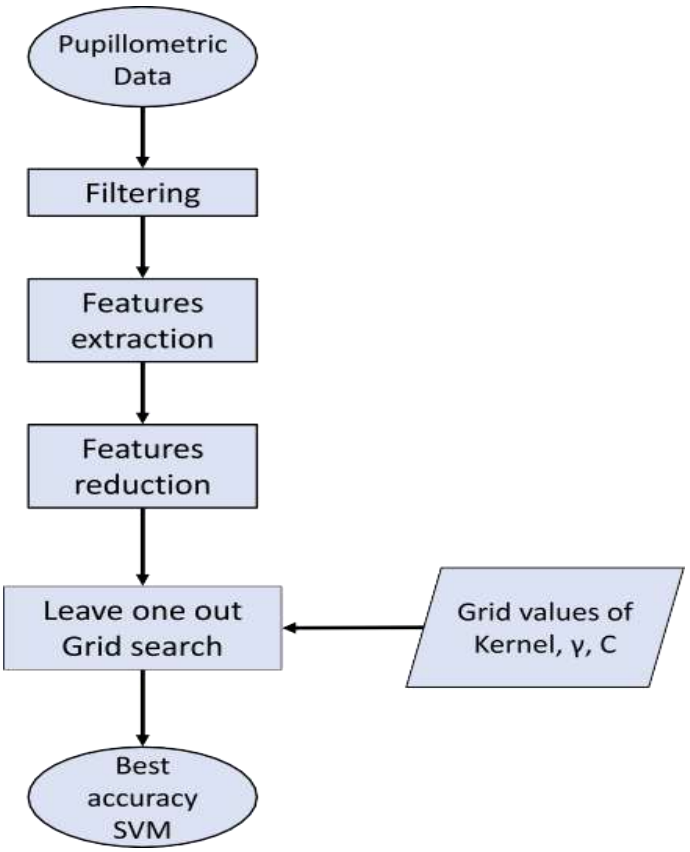

Fig :1 Workflow

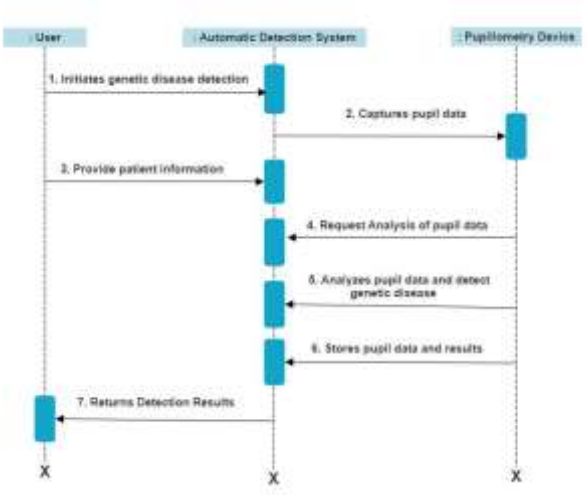

Fig:2 Sequence Diagram

IV. PROPOSED SYSTEM

The proposed methodology introduces an automated disease detection system that leverages pupillometry data and machine learning to enhance diagnostic accuracy. This system is designed to overcome the limitations of traditional clinical evaluations by providing a non-invasive, efficient, and scalable solution for disease detection. The methodology involves a multi-faceted machine learning pipeline that integrates data preprocessing, feature extraction, and classification modeling. Key pupillary response characteristics such as maximum and minimum pupil diameter, delta, channel height (CH), latency, and mean change velocity (MCV) are extracted to capture essential patterns

associated with various disease states. The extracted features are then fed into multiple machine learning models, including Support Vector Machines (SVM), ensemble methods, Extreme Learning Machines (ELM), Multi-Layer Perceptrons (MLP), Random Forests, and a novel hybrid model that combines the strengths of multiple algorithms. This comprehensive approach enhances classification accuracy and ensures robustness in disease detection. By automating the diagnostic process, the system aims to support early detection, minimize reliance on invasive procedures, and improve overall patient outcomes.

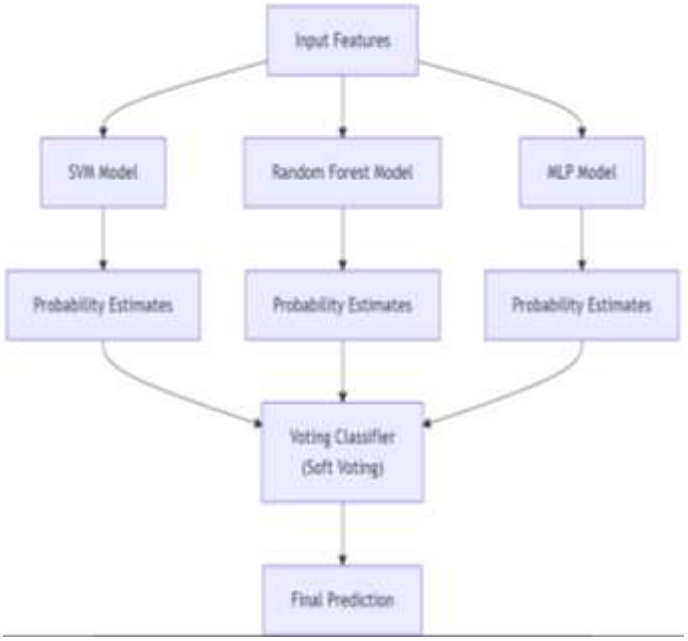

Fig 3:Model Architecture

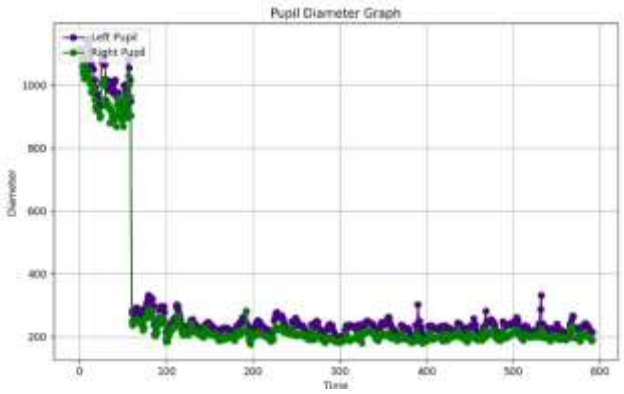

Fig: 4 Pupil Diameter Graph

A. *Support Vector Machine:*

The Support Vector Machine (SVM) model is implemented separately for both left and right eye data, utilizing a radial basis function (RBF) kernel to transform the input features into a higher-dimensional space where linear separation is achievable. This kernel function effectively captures non-linear relationships within the pupillometry data, which is crucial for distinguishing subtle variations indicative of disease. SVM's strength lies in its ability to handle high-dimensional data while maintaining robustness against overfitting, making it a suitable choice for this application, particularly when dealing with complex feature spaces and relatively small datasets. To address class imbalance, the class-weight parameter is adjusted, ensuring that the model maintains both sensitivity and specificity in disease classification.

B. *Ensemble:*

The ensemble model enhances predictive performance by combining the outputs of two SVM classifiers—one for each eye—using a hard voting strategy. This approach leverages the complementary information provided by both eyes, potentially increasing overall classification accuracy and robustness. By aggregating predictions from multiple classifiers, the ensemble model reduces individual biases and variances, improving generalization to new data. This strategy ensures more stable and reliable predictions, particularly when dealing with noisy or complex pupillometry datasets.

C. *Extreme Learning Machine:*

The Extreme Learning Machine (ELM) model offers a computationally efficient alternative for disease classification by utilizing a single hidden layer with randomly assigned weights and biases. The activation function, a hyperbolic tangent (tanh), introduces non-linearity, allowing the model to capture complex patterns in the pupillometry data. ELM is particularly advantageous due to its rapid training speed and potential for high accuracy, making it suitable for real-time applications. By optimizing output weights through a least-squares solution, ELM efficiently maps input features to target disease classes, ensuring fast and effective classification.

D. *Multi Layer Perceptron:*

The Multi-Layer Perceptron (MLP), a type of artificial neural network, employs multiple hidden layers to learn intricate relationships within pupillometry data. The ReLU activation function and an adaptive learning rate enable MLP to capture subtle patterns that traditional models might overlook. Through iterative weight and bias adjustments using backpropagation, the MLP optimizes its performance, making it a powerful tool for disease classification. The hierarchical learning capability of MLP allows it to extract deeper feature representations, which is essential for understanding the complex dynamics of pupillary responses.

E. *Random Forest:*

The Random Forest model, an ensemble of decision trees, provides a robust and versatile approach to disease classification. By aggregating predictions from multiple trees, the model minimizes the risk of overfitting and enhances generalization performance. Bootstrapping and random feature selection introduce diversity among trees, further improving accuracy. Additionally, Random Forest inherently ranks feature importance, making it a valuable tool for identifying the most relevant pupillometry features in disease detection. Its ability to handle high-dimensional datasets efficiently contributes to its effectiveness in this application.

V. DATASET

The dataset used for training and evaluating the proposed hybrid model architecture is carefully curated to ensure comprehensive

representation of disease-specific pupillary response patterns. The hybrid model integrates three advanced neural network paradigms—Convolutional Neural Networks (CNNs), Graph Neural Networks (GNNs), and Transformers—to enhance classification accuracy.

- **CNNs** extract spatial features from pupillometry images, capturing critical structural variations in pupil response.
- **GNNs** model the relational dependencies between extracted features, leveraging graph-based representations to identify complex interactions.
- **Transformers** analyze sequential patterns in pupillary responses, capturing long-range dependencies and improving model robustness.

By combining these methodologies, the dataset is effectively leveraged to distinguish between normal and disease-affected pupillary behavior, enhancing the reliability of automated disease detection.

VI. TESTING AND VALIDATION

Fig:5 Dashboard

This is the main interface of PulmoDetect, introducing its AI-powered system for early genetic disease detection using pupillometry. It outlines the simple 3-step process: Prepare Data, Upload File, and View Results. The platform emphasizes high accuracy and secure analysis for clinical use.

Fig :6 Results

This dashboard shows an overview of the total samples analyzed, the number of positive and negative cases, and their respective percentages. It enables users to understand the distribution of results at a glance. A "New Analysis" button allows for fresh uploads and testing.

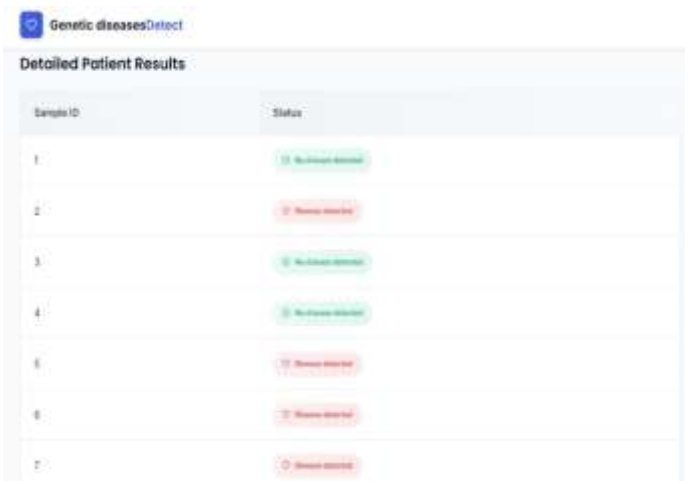

Fig : 7 Detailed Results

This screen displays the individual diagnostic outcomes for each patient sample. Each row indicates whether a genetic disease was detected or not. It provides a clear, color-coded summary for quick analysis.

VII. RESULTS

Evaluation Metrics	Score
Existing Accuracy	0.86
Proposed Accuracy	0.936

VIII. CONCLUSION

This study has demonstrated the potential of machine learning in analyzing pupillometry data for automated disease detection. By carefully extracting key features from pupillary responses and systematically testing various models—including SVMs, ensemble methods, ELM, MLPs, Random Forests, and a hybrid approach—we have shown that accurate disease classification is possible. The strong performance of the hybrid model highlights the advantages of integrating multiple algorithms to improve diagnostic accuracy. Our findings reinforce the value of pupillometry as a non-invasive tool for medical diagnosis and lay a solid foundation for its future clinical use. Ultimately, this research paves the way for earlier disease detection, more efficient screening processes, and better patient outcomes.

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