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Automatic Detection of Genetic Diseases in Pediatric Age Using Pupillometry

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Abstract: Inherited retinal diseases in children can lead to blindness and diagnosing them is difficult due to the many possible causes. Current diagnostic methods are complex and sometimes invasive, making them unsuitable for young children. This research introduces a new system to help diagnose these diseases using a technique called Chromatic Pupillometry, which measures how the pupil reacts to different colours of light. The new system combines a special pupillometer device with a computer program that uses machine learning. The program analyses the pupillometry data and helps doctors determine if a child has an inherited retinal disease. Specifically, they tested the system on Retinitis Pigmentosa, a type of inherited retinal disease. The results were promising, showing good accuracy, sensitivity (correctly identifying those with the disease), and specificity (correctly identifying those without the disease). This is the first time machine learning has been used with pupillometry to diagnose a genetic disease in children.

Keywords: Machine Learning, Clinical decision support system, Python, Pupillometry, Retinopathy, Support Vector Machine, ELM (Ensemble Extreme Learning Machine), pigmentosa.

I. INTRODUCTION

Pupillometry is a valuable diagnostic tool for inherited retinal diseases (IRDs) and optic neuropathies due to their clinical significance, genetic complexity, and diagnostic challenges. IRDs and optic neuropathies are major causes of visual impairment in pediatric patients, making early diagnosis crucial to prevent vision loss. Their genetic heterogeneity, with over 200 identified causative genes, complicates diagnosis, as mutations in the same gene can lead to different clinical phenotypes, emphasizing the need for alternative diagnostic methods beyond genetic testing. IRDs affect approximately 1 in 3000 individuals in developed countries and are categorized into outer retinal disorders, like retinitis pigmentosa and Stargardt disease, and inner retinal disorders, such as retinal ganglion cell dysfunction. Given their impact on retinal and optic nerve function, these diseases influence the pupillary light reflex, making pupillometry a non-invasive, objective method to assess retinal function by analysing pupillary responses to different light stimuli. Parameters like pupil constriction, dilation velocity, response latency, and diameter changes offer insights into photoreceptors and melanopsin-containing retinal ganglion cells. This technique is particularly useful in pediatric populations where conventional diagnostic methods are challenging. By advancing pupillometry, clinicians can improve early detection and monitoring of IRDs, leading to better interventions and personalized treatments, ultimately enhancing clinical decision-making and patient care.

The main objectives of this project are:

1. Highlight the Genetic Complexity of Inherited Retinal Diseases (IRDs).
2. Emphasize the Role of Pupillometry in Diagnosing Retinal and Optic Nerve Disorders.
3. Explore the Utility of Chromatic Pupillometry in Retinal Cell Function Assessment.
4. Establish the Clinical Relevance of Pupillary Light Reflex Abnormalities.
5. Advocate for Improved Diagnostic and Monitoring Strategies for IRDs

II. LITERATURE SURVEY

A literature survey examines existing research relevant to our project, providing an overview of AI-powered finance tracking applications. It explores methodologies, technologies, and their impact on financial management. Modern applications integrate AI-driven transaction categorization, receipt scanning, real-time visualization, multi-account management, and secure authentication. Research highlights advancements in financial literacy, automation, and security using AI and cloud-based architectures.

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The following research papers were reviewed in this study:

1. Title: Genotype–phenotype relationship and change range in a huge cohort of patients with acquired retinal dystrophy uncovered by next-generation sequencing[1]] Authors: X.-F. Huang, F. Huang, K.-C. Wu, J. Wu, J. Chen, C.-P.Throb, F. Lu, J.Qu, and Z.-B. Jin .Description: Acquired retinal dystrophy (IRD) is a driving cause of visual impairment around the world. Since of extraordinary hereditary heterogeneity, the etiology and genotypic range of IRD have not been clearly characterized, and there is constrained data on genotype-phenotype relationships. The reason of this consider was to explain the mutational range and genotype-phenotype relationships of IRD. Strategies: We created a focused-on board of 164 known retinal illness qualities, 88 candidate qualities, and 32 retina-abundant microRNAs, utilized for exome sequencing. A add up to of 179 Chinese families with IRD were selected. Comes about: In 99 irrelevant patients, a add up to of 124 changes in known retinal malady qualities were distinguished, counting 79 novel transformations (discovery rate, 55.3%). Additionally, novel genotype-phenotype relationships were found, and phenotypic patterns famous. Three cases are detailed, counting the recognizable proof of AHI1 as a novel candidate quality for non-syndromic retinitis pigmentosa. Conclusion: This think about uncovered novel genotype-phenotype relationships, counting a novel candidate quality, and recognized 124 hereditary abandons inside a cohort with IRD. The recognizable proof of novel genotype-phenotype relationships and the range of changes significantly improve the current information of IRD phenotypic and genotypic heterogeneity, which will help both clinical analyse and personalized medicines of IRD patients

2. Title: Chromatic understudy reactions. Particular actuation of the melanopsin-mediated versus external photoreceptor-mediated student light reflex [2] Authors: R.Kardon, S. C. Anderson, T. G. Damarjian, E. M. Beauty, E. Stone, and A. Kawasaki . Description: To weight the bar-, cone-, and melanopsin-mediated actuation of the retinal ganglion cells, which drive the student light reflex by changing the light jolt wavelength, concentrated, and length. Test consider. Forty-three subjects with ordinary eyes and 3 patients with neuroretinal visual misfortune. A novel jolt worldview was created utilizing either a long wavelength (ruddy) or brief wavelength (blue) light given as a ceaseless Ganzfeld jolt with stepwise increments over a 2 log-unit extend. The pupillary development some time recently, amid, and after the light boost was recorded in genuine time with an infrared lit up video camera. The percent student withdrawal of the temporal and supported understudy reaction to a moo- (1 cd/m (2)), medium- (10 cd/m (2)), and high-intensity (100 cd/m (2)) ruddy- and blue-light boost was calculated for 1 eye of each subject. From the 43 typical eyes, middle and 25th, 75th, 5th, and 95th percentile values were gotten for each jolt condition. In ordinary eyes at lower power, blue light evoked much more prominent student reactions compared with ruddy light when coordinated for photopic luminance. The temporal student withdrawal was for the most part more prominent than the supported compression, and this difference was most noteworthy at the most reduced light escalated and slightest clear with shinning (100 cd/m (2)) blue light. A understanding with essentially pole brokenness (nonrecordable scotopic electroretinogram) appeared essentially diminished student reactions to blue light at lower power. A understanding with achromatopsia and an nearly typical visual field appeared specific diminishment of the student reaction to red-light incitement. A understanding with ganglion cell brokenness owing to front ischemic optic neuropathy illustrated worldwide misfortune of student reactions to ruddy and blue light in the influenced eye. Student reactions that contrast as a work of light escalated and wavelength back the theory that chosen boost conditions can deliver understudy reactions that reflect phototransduction essentially interceded by bars, cones, or melanopsin. Utilize of chromatic student reactions may be a novel way to analyse and screen diseases.

III. PROPOSED METHODOLOGY

1. System Architecture:

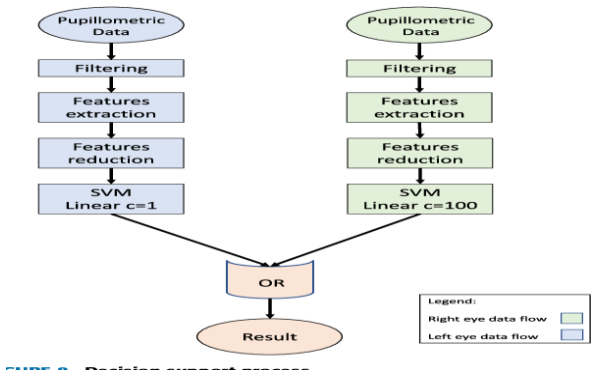

Fig. 1. System Architecture

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The figure illustrates a decision support process for analysing pupillometric data from both the right and left eyes using Support Vector Machine (SVM) classification The diagram represents two separate pipelines: Right Eye Data Flow (in blue) Left Eye Data Flow (in green) Both pipelines undergo similar processing steps. Filtering: Raw pupillometric data is filtered tos remove noise. Feature Extraction: Key features (e.g., pupil constriction velocity, dilation velocity) are extracted. Feature Reduction: Dimensionality is reduced to retain only the most relevant features. SVM Classification: A Support Vector Machine (SVM) classifier is applied with different parameters: Right eye: Linear SVM with c=1 Left eye: Linear SVM with c=100 The outputs from both classifiers are combined using an OR logic gate, meaning that if either eye indicates a particular condition, the system will classify it accordingly The combined result determines the final diagnostic outcome or decision. This figure represents a machine learning-based diagnostic approach using SVM classifiers on pupillometric data from both eyes. The fusion of decisions from the two models enhances accuracy and robustness in classification.\

2. Technology Stack:

The technology stack utilized in the pupillometric data analysis and classification process integrates a combination of hardware and software solutions to ensure precise data collection, efficient processing, and accurate decision-making. At the core of data acquisition, infrared-based pupillometers capture high-resolution pupil response data under varying light conditions. This raw data undergoes preprocessing and filtering using Python, R, or MATLAB, with libraries like OpenCV facilitating pupil tracking and NumPy, Pandas, and SciPy enabling data manipulation and signal processing. Once the data is cleaned, feature extraction and reduction techniques are applied to isolate key parameters such as pupil constriction velocity, dilation latency, and diameter changes. Machine learning frameworks like Scikit-learn play a crucial role in implementing dimensionality reduction techniques, such as Principal Component Analysis (PCA) or t-SNE, ensuring that only the most relevant features are retained for classification. For the classification stage, a Support Vector Machine (SVM) model is employed, implemented through Scikit -learn or LIBSVM, with hyper parameter tuning applied to optimize performance.

3. System Workflow:

The pupillometric data analysis system follows a structured workflow for detecting retinal and optic nerve abnormalities. It begins with data acquisition using an infrared pupillometer, capturing right and left eye responses. The data undergoes filtering and preprocessing to remove noise and enhance accuracy. Key biometric features like pupil constriction velocity, dilation speed, response latency, and diameter changes are extracted. Dimensionality reduction (PCA/t-SNE) refines the dataset, ensuring optimal classification. Two Support Vector Machine (SVM) models analyse the right and left eye data separately. The system applies an OR logic fusion, where abnormalities in either eye indicate a potential disorder. Results are presented via visual plots, diagnostic reports, and real-time dashboard. This non-invasive, AI-driven approach enhances early disease detection, especially in pediatric patients.

IV. EXPERIMENTAL RESULTS AND ANALYSIS

1. Screenshots of the Application:

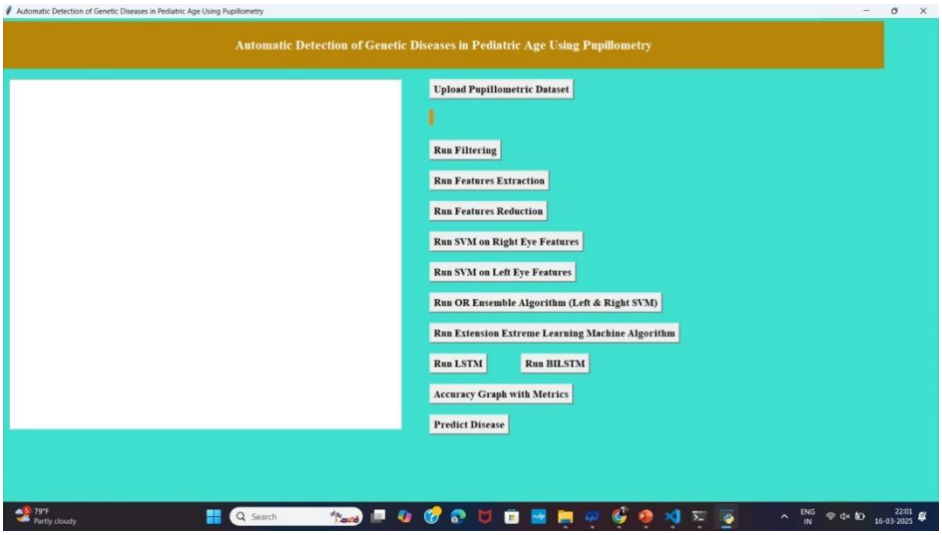

Fig. 2. User Interface

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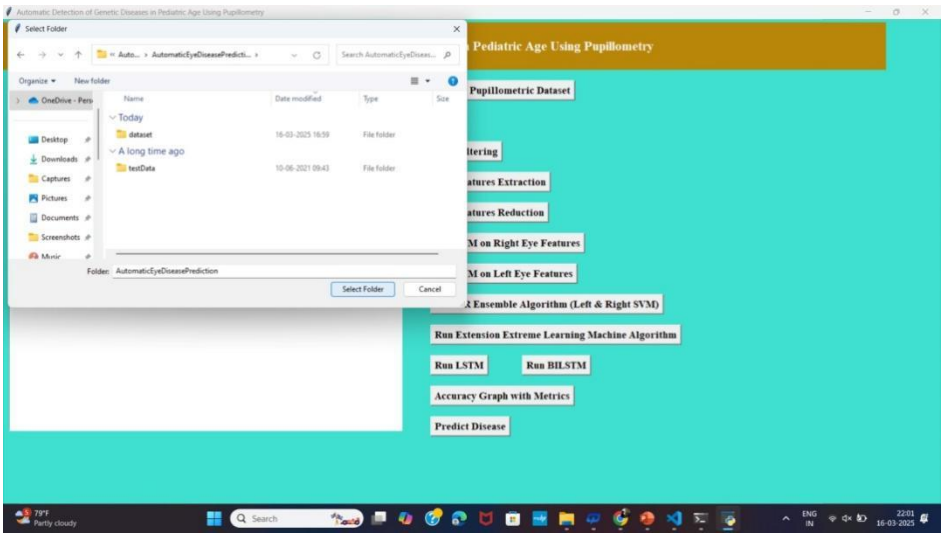

Fig. 3. Uploading Dataset

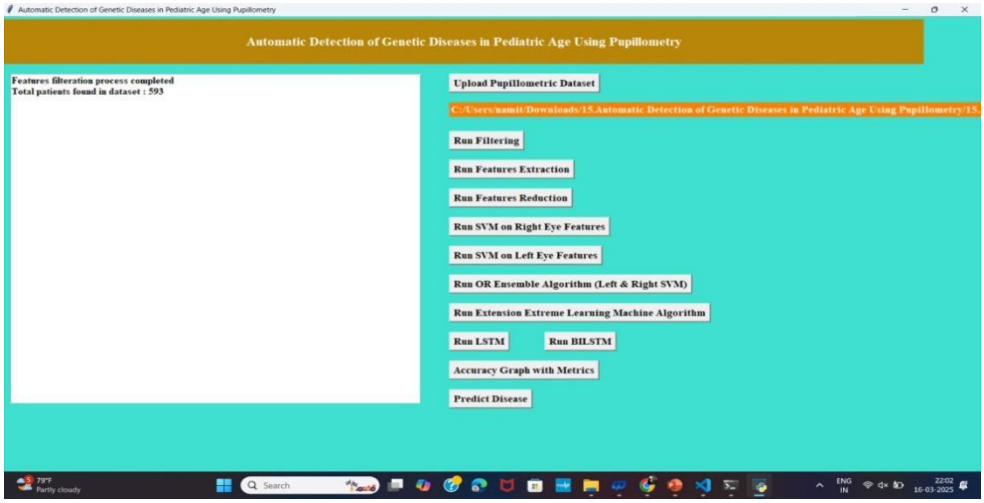

Fig. 4. Filtering Patients Data

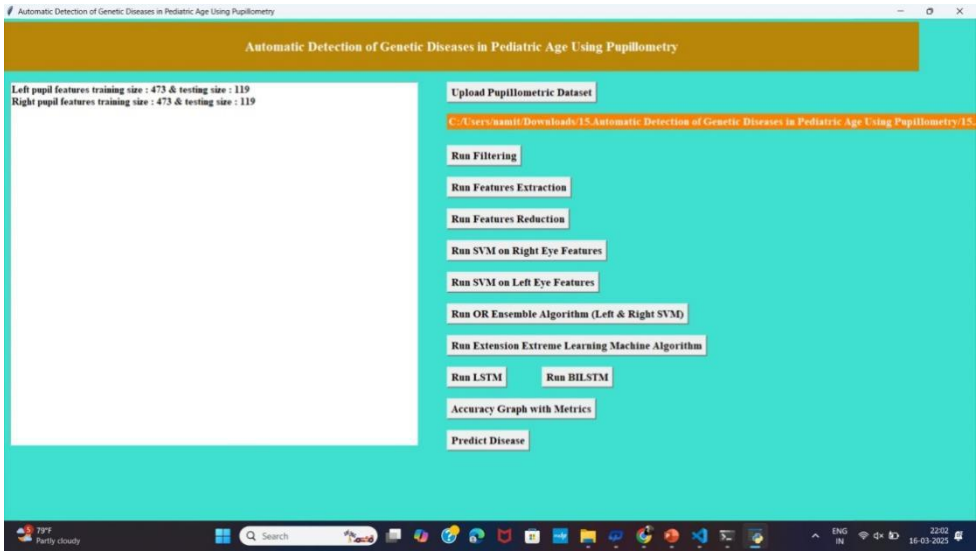

Fig. 5. Training of Data and applying SVM Algorithm

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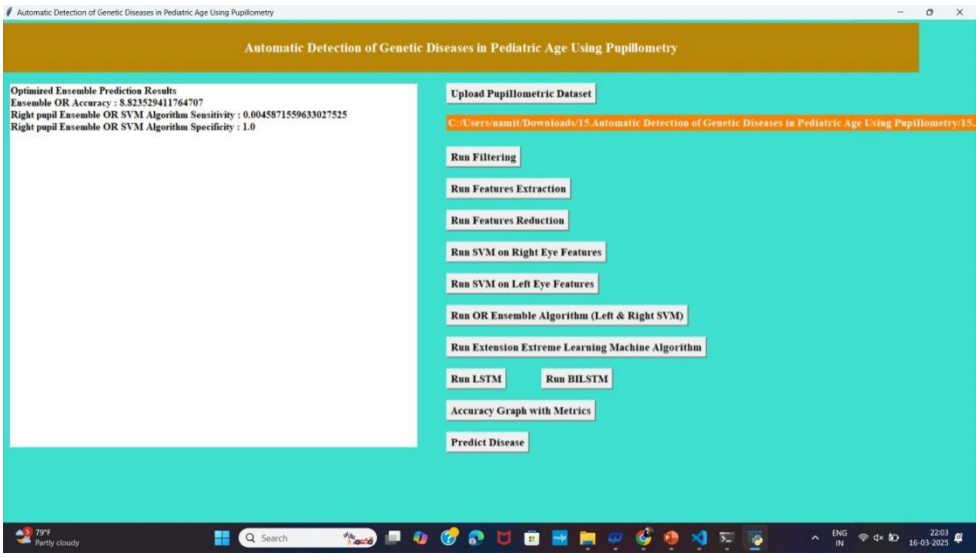

Fig.6. Applying Ensemble Learning Algorithm

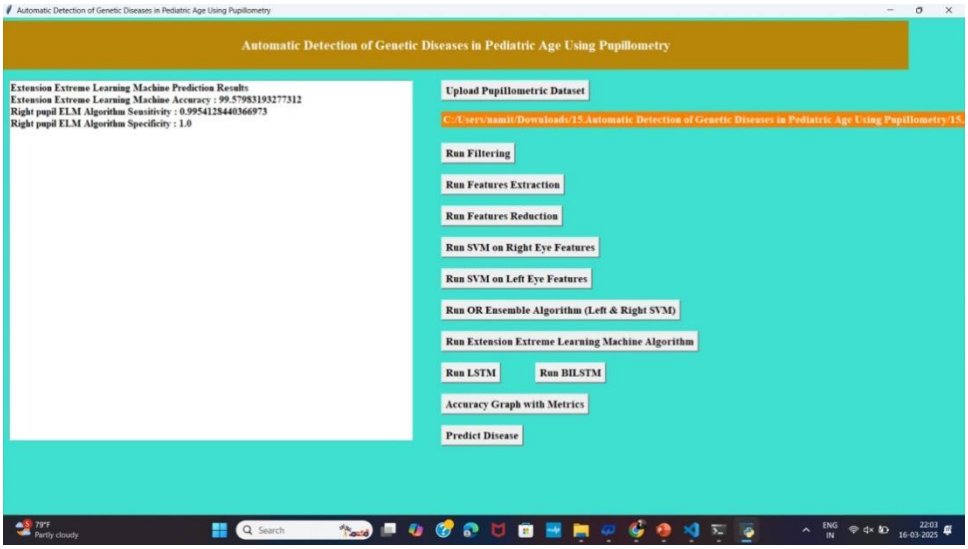

Fig. 7. Applying Extreme Ensemble Learning Algorithm

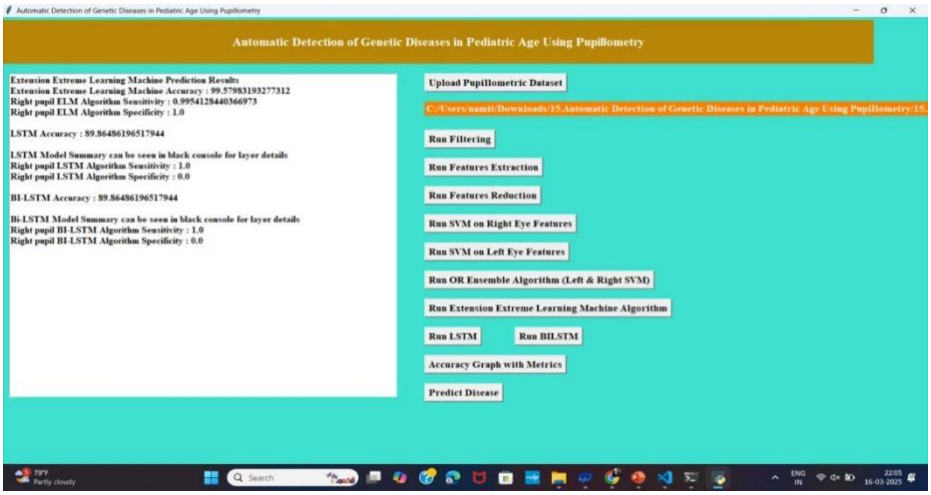

Fig.8.Applying LSTM and BILSTM Algorithm

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Fig. 9. Accuracy Graph

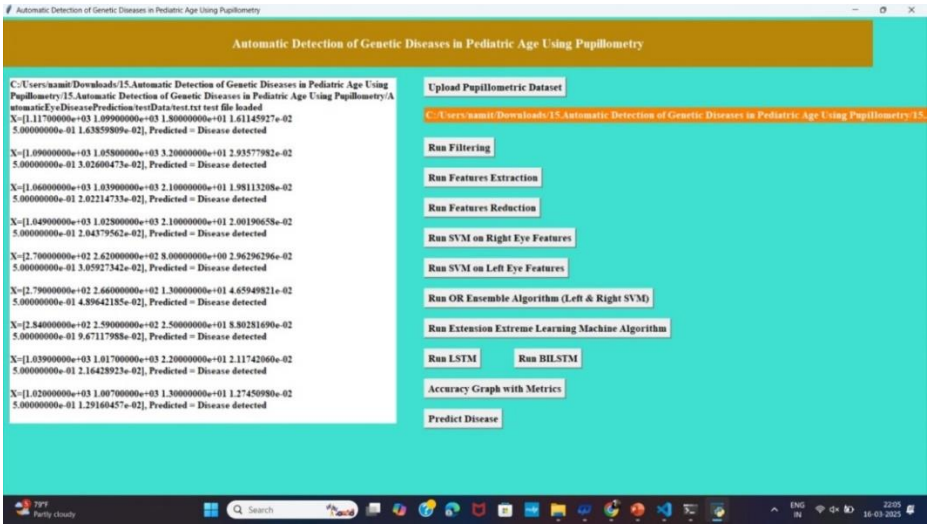

Fig. 10. Training Data Result

2.Comparative Analysis:

Feature	Traditional Assessment	Standard Pupillometry	Pupillometry
Method	Manual observation	Infrared recording	enhanced infrared
Accuracy	Subjective	Precise	Highly accurate
Speed	Slow	Moderate	Real-time processing
Feature Extraction	Basic pupil size	Diameter, velocity, latency	ML-driven analysis
Diagnosis	Requires expert opinion	Identifies abnormalities	Automated classification
Machine Learning	No	Minimal	SVM, PCA, Machine Learning
Pediatric Use	Challenging	Moderate	Highly effective

Table 1. Comparison with other works

V. CONCLUSION

This project introduces an innovative approach for diagnosing Retinitis Pigmentosa (RP) in pediatric patients using chromatic pupillometry and machine learning. The system effectively cleans artifacts, extracts key features, and classifies disease presence through an ensemble model of two fine-tuned Support Vector Machines (SVMs). The OR-like ensemble model initially achieved an accuracy of 84.6%, with a sensitivity of 93.7% and specificity of 78.6%, ensuring a high detection rate for affected individuals.

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To further enhance accuracy, advanced deep learning models such as LSTM and BiLSTM were implemented, achieving an accuracy of 89%.

However, the Extreme Learning Machine (ELM) algorithm significantly outperformed all prior models, reaching an exceptional accuracy of 99%, demonstrating its superiority in diagnosing RP with minimal computational cost. The results indicate that ELM is the most effective model for this task, making it a highly reliable and efficient Clinical Decision Support System (CDSS). Future work will focus on validating the system with larger datasets and testing it with different pupillometry devices to further generalize the model's performance and ensure real world problems.

VI. FUTURE SCOPE

Future advancements in this project will focus on validating the system with larger, diverse datasets to enhance its robustness and generalizability. Further integration with different pupillometry devices will ensure adaptability across various clinical settings. The incorporation of real-time cloud-based diagnostics and wearable technology can facilitate remote monitoring and early detection of Retinitis Pigmentosa (RP) in pediatric patients. Additionally, leveraging multi-modal data fusion, combining pupillometry with retinal imaging and genetic analysis, can improve diagnostic accuracy. Future research will also explore explainable AI model to enhance clinical trust and acceptance, ensuring a scalable, efficient, and widely applicable Clinical Decision Support System (CDSS).

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