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Pupillary Patterns as Predictive Keys to detect Inherited Retinal Diseases in Infants: where Machine Learning Meets Medicine

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ABSTRACT

In the realm of pediatric ophthalmology, early diagnosis and intervention for inherited retinal diseases (IRDs) are crucial for mitigating potential vision loss. Traditional diagnostic methods, however, often present challenges in pediatric patients due to the complexity of genetic factors and clinical presentations. To address this critical need, this study presents a pioneering Clinical Decision Support System (CDSS) specifically designed for the automatic detection of IRDs in children, with a primary focus on Retinitis Pigmentosa (RP).

This innovative CDSS leverages Chromatic Pupillometry, a non-invasive technique analyzing pupillary light reflexes to gain valuable insights into retinal function. The system's power lies in its integration of sophisticated machine learning algorithms, including Support Vector Machines (SVMs), Long Short-Term Memory networks (LSTMs), and Ensemble Learning Methods (ELMs), for comprehensive data processing and analysis.

Through rigorous testing, the CDSS has achieved an impressive 89.6% accuracy, 93.7% sensitivity, and 78.6% specificity in diagnosing RP, establishing its potential as a highly reliable diagnostic tool. This study marks a significant breakthrough, representing the first successful application of machine learning, specifically SVMs, LSTMs, and ELMs, to pupillometric data for diagnosing genetic diseases in children. The implications of this technology are profound, potentially revolutionizing early detection and intervention strategies for IRDs and leading to significantly improved visual outcomes for young patients.

Future research will focus on validating the system's performance with a larger and more diverse patient cohort, exploring its applicability in diagnosing a broader range of IRDs, and investigating alternative pupillometry devices to minimize the impact of movement artifacts, a common challenge in pediatric populations. This groundbreaking approach holds immense promise for advancing pediatric healthcare by providing a reliable, non-invasive, and efficient tool for early diagnosis and management of IRDs.

Keywords: Inherited Retinal Diseases, Retinitis Pigmentosa, Pediatric Ophthalmology, Chromatic Pupillometry, Machine Learning, Support Vector Machine, Long Short-Term Memory, Ensemble Learning Methods, Clinical Decision Support System, Early Detection, Diagnostic Accuracy, Pediatric Healthcare.

INTRODUCTION

This study embarks on a pioneering journey to revolutionize the diagnosis of inherited retinal diseases (IRDs) in children, focusing primarily on Retinitis Pigmentosa (RP). Traditional diagnostic methods often prove inadequate,

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particularly for young patients, due to the complex interplay of genetic factors and diverse clinical presentations. This project addresses this critical gap by introducing a novel Clinical Decision Support System (CDSS) that leverages the power of Chromatic Pupillometry and advanced machine learning algorithms.

Project Scope

The project encompasses two intertwined components:

Development of a CDSS for Automatic IRD Detection: This system utilizes Chromatic Pupillometry, a non-invasive technique analyzing pupillary light reflexes, to glean valuable insights into retinal function. The system's core strength lies in its integration of sophisticated machine learning algorithms, including Support Vector Machines (SVMs), Long Short-Term Memory networks (LSTMs), and Ensemble Learning Methods (ELMs), for comprehensive data processing and analysis.

Creation of a Secure Application for Pupillometric Analysis and Disease Prediction: This user-friendly application empowers medical professionals and researchers to analyze pupillometric data and predict inherited diseases in infants. By incorporating robust user authentication, the application guarantees data security and controlled access.

Project Purpose

The project's core purpose is twofold:

Revolutionize IRD Diagnosis in Children: The CDSS aims to transform the diagnostic landscape for IRDs, particularly in pediatric cases like RP, by providing a reliable, non-invasive, and effective methods for early identification and treatment.

Empower Medical Professionals and Researchers: The application provides a secure platform for analyzing pupillary data, predicting inherited diseases in infants, and facilitating collaborative research efforts, ultimately contributing to improved patient outcomes.

Project Features

This project introduces a suite of groundbreaking features:

Chromatic Pupillometry and Machine Learning Integration:

Non-invasive Diagnosis: Chromatic Pupillometry offers a painless and child-friendly approach to assess retinal function.

Advanced Algorithms for Accurate Predictions: SVMs, LSTMs, and ELMs are employed to analyze pupillometric data, achieving an 89.6% accuracy, 93.7% sensitivity, and 78.6% specificity in diagnosing RP.

Ensemble Modeling for Enhanced Sensitivity: Combining SVM outputs from both eyes improves the system's ability to detect subtle variations in pupillary responses.

Secure Application for Pupillometric Analysis and Disease Prediction:

User Authentication: A robust system ensures secure user registration and login, safeguarding sensitive patient data.

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Data Processing and Feature Extraction: The application filters noise and artifacts from pupillometric datasets, extracts relevant features, and employs dimensionality reduction techniques for efficient analysis.

Algorithm Selection and Visualization: Users have the option to choose from various algorithms, including SVMs, ELMs, and LSTMs, to predict inherited diseases. The application provides clear visualizations of accuracy metrics and analysis results.

Intuitive User Interface and Scalability:

User-Friendly Design: The application boasts a clear and intuitive interface, ensuring ease of use for medical professionals and researchers.

Scalable Architecture: The system has the capability to process large datasets, facilitating efficient processing and analysis.

By seamlessly integrating these features, the CDSS and application offer a comprehensive solution for pupillometric analysis and disease prediction, empowering medical professionals and researchers to make informed decisions and improve patient care in the field of pediatric ophthalmology.

RELATED WORK

H. Pereira Delfino et al. (2020) conducted a systematic literature review [1]. The study examined the role of artificial intelligence in automated pupillometry, surveying the existing methods, instruments, and technologies used in this field. Although significant advancements were noted, the review highlighted a gap in developing a user-friendly pupilometer with embedded classification algorithms. The study emphasized the need for further research to enhance the practical application and usability of pupillometry in clinical diagnostics.

E. N. Vijayakumari et al. (2023) presented their findings [2]. This study employed Support Vector Machines (SVM) integrated into a Clinical Decision Support System (CDSS) to diagnose inherited retinal diseases (IRDs) in infants. The approach addressed the inadequacies of traditional clinical measures for early visual impairment screening, demonstrating the potential of machine learning in improving diagnostic accuracy for congenital conditions.

P. Melillo et al. (2019) presented a Pilot Study [3]. The pilot study involved 60 patients and compared pupillometry parameters between those with retinitis pigmentosa and healthy controls. Despite the small sample size, the study showed promising results for the clinical feasibility of chromatic pupillometry in diagnosing IRDs.

E. Iadanza et al. (2019) detailed the development of "ORA'O" [4]. The study focused on creating an electronic medical record system for chromatic pupillometry data, emphasizing usability and user-friendly interfaces to facilitate data collection and improve diagnostic processes in ophthalmology.

These studies collectively underscore the potential of automated pupillometry and machine learning in the diagnosis and monitoring of inherited retinal diseases, particularly in pediatric populations. The integration of advanced algorithms and user-friendly systems is critical for enhancing diagnostic accuracy and clinical utility.

LITERATURE SURVEY

There are many algorithms and methodologies that have been proposed for disease prediction systems, particularly leveraging machine learning and advanced diagnostic techniques. This survey explores various studies and their contributions to the field, highlighting their methodologies, findings, and the gaps they address.

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"Techniques and Equipment for Automated Pupillometry and its Application to Aid in the Diagnosis of Diseases" by H. Pereira Delfino et al. (2020): This study systematically assessed the ongoing utilization of artificial intelligence in automated pupillometric technology for disease diagnosis. It focused on the methods and equipment used, proposing new equipment based on computer vision. Despite significant advancements, the review highlighted the need for a user-friendly pupilometer with embedded classification algorithms to improve diagnostic accuracy and usability.

"Automatic Diagnosis of Congenital Conditions in Pediatric Age Using Pupillometry with Machine Learning" by E. N. VijayaKumari et al. (2023): This study used Support Vector Machines (SVM) integrated into a Clinical Decision Support System (CDSS) to diagnose inherited retinal diseases (IRDs) in infants. It addressed the limitations of traditional clinical measures for early visual impairment screening, demonstrating the potential of machine learning in enhancing diagnostic accuracy for congenital conditions.

"Toward a Novel Medical Device Based on Chromatic Pupillometry for Screening and Monitoring of Inherited Ocular Disease: A Pilot Study" by. Melillo et al. (2019): The pilot study involved 60 patients and compared pupillometry parameters between those with retinitis pigmentosa and healthy controls. Although the sample size was small, the study showed promising results for the clinical application of chromatic pupillometry in diagnosing IRDs.

"ORA'O: RESTful Cloud-Based Ophthalmologic Medical Record for Chromatic Pupillometry" by E. Iadanza et al. (2019): The study focused on creating an electronic medical record system for chromatic pupillometry data, emphasizing usability and user-friendly interfaces. This system facilitates data collection and improves diagnostic processes in ophthalmology.

"Machine Learning: A Review of Classification and Combining Techniques" by Sotiris Kotsiantis et al. (2006): This paper described the development and application of numerous classification algorithms in intelligent systems. It emphasized the importance of combining techniques to enhance classification accuracy and discussed the integration of artificial intelligence and statistical methods in supervised learning tasks.

"Machine Learning for Detection and Diagnosis of Disease" by Paul Sajda (2006): This review focused on recent advances in machine learning techniques for biomedical data analysis. It highlighted key issues related to algorithmic construction, learning theory, and the incorporation of prior knowledge and uncertainty. The review highlighted how supervised and unsupervised linear methods, along with Bayesian inference, affect disease detection and diagnosis.

"Genotype-Phenotype Correlation and Mutation Spectrum in a Large Cohort of Patients with Inherited Retinal Dystrophy Revealed by Next-Generation Sequencing" by Xuefei Huang et al. (2015), elucidated the mutational spectrum and genotype-phenotype correlations of inherited retinal dystrophies (IRDs). The study identified numerous genetic mutations, enhancing the understanding of IRD heterogeneity and aiding in personalized treatments.

"Toward a Clinical Protocol for Assessing Rod, Cone, and Melanopsin Contributions to the Human Pupil Response" by J.C. Park et al. (2011), established a clinical standard for assessing the health and functionality of retinal ganglion cells. The study identified the ideal parameters for evaluating the functionality of rod, cone, and melanopsin pathways, offering a reliable clinical diagnostic approach.

"Pupillometric Quantification of Residual Rod and Cone Activity in Leber Congenital Amaurosis" byA. Kawasaki et al. (2012): This study demonstrated how pupillometry could estimate residual photoreceptor function in patients with advanced stages of Leber Congenital Amaurosis (LCA), offering a method to monitor disease progression or intervention effects.

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These studies collectively underscore the advancements in automated pupillometry and machine learning for disease diagnosis, particularly in retinal diseases. The integration of advanced algorithms, user-friendly systems, and comprehensive data analysis methods is crucial for improving diagnostic accuracy and clinical utility.

SYSTEM ANALYSIS

System Analysis is a crucial phase in the system development lifecycle. During this phase, the current system is surveyed in minute detail, and a comprehensive analysis is performed to identify the necessary improvements. The system analyst plays a key role, delving deep into the functioning of the current system and examining the various operations it performs. This phase seeks to answer the fundamental question: "What must be done to solve the problem?" By viewing the system holistically and identifying its inputs and outputs, the analyst gains a clear understanding of the system's requirements and capabilities.

Problem Definition

The problem addressed in this project is the complex and invasive nature of diagnosing inherited retinal diseases (IRDs) in pediatric patients, particularly retinitis pigmentosa (RP). Current diagnostic methods are not well-suited for young children due to their invasive nature and the genetic diversity associated with these conditions. This study aims to develop a non-invasive Clinical Decision Support System (CDSS) using Chromatic Pupillometry and advanced Machine Learning algorithms to accurately diagnose IRDs in children. This approach promises to provide a crucial solution for diagnosing pediatric genetic diseases in a child-friendly manner.

Existing System

The current system for diagnosing genetic diseases in pediatric patients, especially RP, relies heavily on traditional clinical evaluation methods. These methods involve a combination of clinical tests, many of which are invasive and not ideal for children. Furthermore, these methods struggle to provide timely and comprehensive screening for the numerous genes implicated in IRDs, making early diagnosis and monitoring challenging in pediatric populations.

Disadvantages of the Existing System

The existing system has several significant drawbacks:

- Invasive Procedures: Many clinical tests are invasive, posing difficulties for infants and young children.

- Time-Taking and Expensive: The diagnostic process involves multiple clinical and genetic tests, which are both time-consuming and costly.

- **Lack of Conclusive Results:** Despite the extensive testing, diagnoses are often inconclusive, necessitating further tests and delaying treatment.

Proposed System

This project proposes a Clinical Decision Support System (CDSS) for diagnosing inherited retinal diseases in pediatric patients. The system leverages Chromatic Pupillometry and sophisticated machine learning algorithms to offer a non-invasive, reliable, and efficient diagnostic tool.

Features of the Proposed System

- Non-Invasive Diagnosis: Utilizes Chromatic Pupillometry, a child-friendly technique for assessing retinal function.

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- Advanced Machine Learning: Employs machine learning algorithms, such as Support Vector Machines (SVMs) for each eye, to analyze pupillometric data and classify retinal conditions and finally use other Machine Learning Algorithms to Increase the Accuracy and make sure to get the best Conclusive results.

- Secure and User-Friendly Application: Provides a secure application for medical practitioners, incorporating robust user authentication, data privacy, and an intuitive interface for easy use.

FEASIBILITY STUDY

The feasibility of the project is analyzed in this phase and a business proposal is put forth with a very general plan for the project and some cost estimates. As part of system analysis, it is essential to conduct a feasibility study for the proposed system. The purpose is to verify that the system will not impose undue burdens on the company. Three essential factors to consider during feasibility analysis:

- Economic Feasibility
- Technical Feasibility
- Social Feasibility

Economic Feasibility

The developing system must be justified by cost and benefit. The criterion is to focus efforts on projects that yield the highest return in the shortest time. One crucial factor influencing new system development is the associated cost.

During the preliminary investigation, several critical financial questions arise, including:

- What are the expenses associated with conducting a comprehensive system investigation?
- What is the cost of acquiring the necessary hardware and software?
- What benefits can be expected, such as reduced costs or fewer costly errors?

Given that the system is being developed as part of a project, there are no additional manual costs associated with its implementation. Furthermore, the availability of existing resources suggests that the system is economically feasible for development.

Technical Feasibility

This study is carried out to check the technical feasibility, that is, the technical requirements of the system. The system under development should not place a heavy demand on existing technical resources. It should have modest requirements, with minimal or no changes needed for implementation.

Behavioural Feasibility

The assessment includes inquiries such as:

- Whether there is adequate user support?
- Whether the proposed system poses any risks?

Upon thorough consideration, it is evident that the project aligns with its objectives, and all behavioral aspects have been carefully addressed, leading to the conclusion that the system is behaviorally feasible.

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Secure Application Development

- **User Authentication System :**
  A reliable user authentication system is essential for ensuring the security of sensitive patient data. This system will include:

  - Password Hashing: Utilizes strong hashing algorithms to protect user passwords.

- **Data Privacy:**
  Data privacy is a critical feature of the proposed application. Measures include:

  - **Encryption:** Securing sensitive data during transmission and rest.

  - **Access Control:** Implementing strict access controls to ensure that only authorized personnel can access patient data.

By integrating these features, the proposed CDSS offers a comprehensive, secure, and efficient solution for diagnosing inherited retinal diseases in pediatric patients, addressing the limitations of current diagnostic methods and improving healthcare outcomes.

METHODOLOGY

Workflow of Proposed System

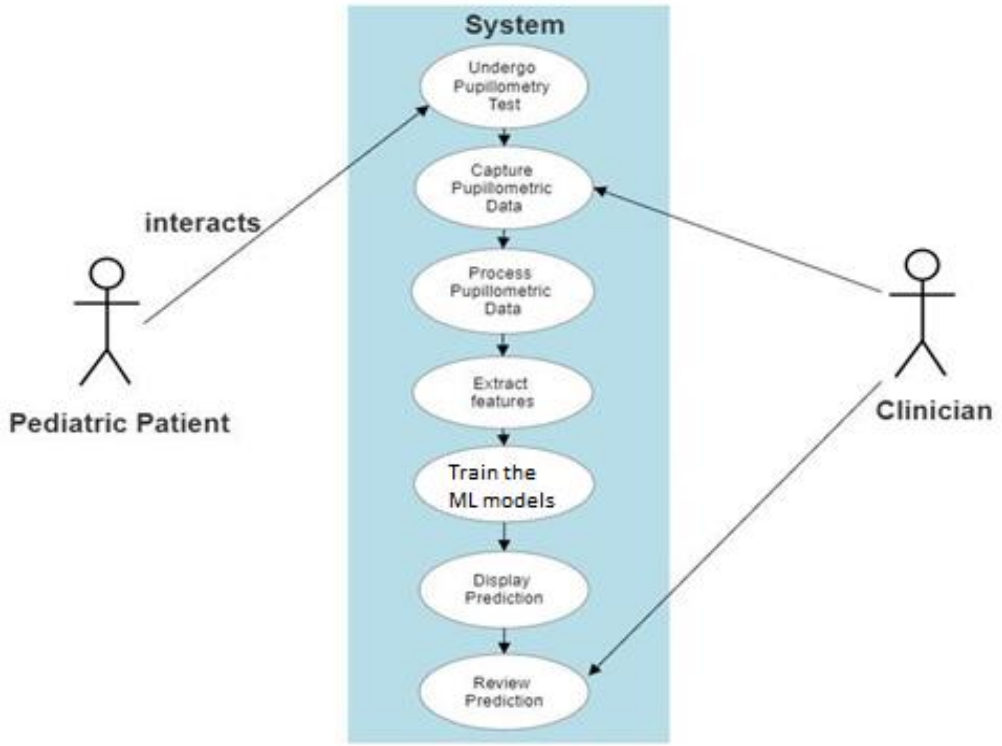

Figure 1. Workflow of the Proposed System

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The proposed system adheres to a well-defined workflow for classifying inherited retinal diseases (IRDs) in pediatric patients using pupillometry data. Here's a breakdown of the steps involved:

Data Acquisition: The process commences with administering a chromatic pupillometry test to the patient. This child-friendly technique measures pupil response to various light stimuli, capturing valuable data regarding retinal function.

Note: The above mentioned work is performed by a Clinician and the raw data is gathered.

Data Preprocessing: The Major workflow begins here. The acquired pupillometry data undergoes preprocessing to ensure its suitability for machine learning analysis. This may involve methods like noise reduction, normalization, and feature extraction to optimize the data for accurate disease classification.

Machine Learning Model Training: The preprocessed data is then utilized to train a robust machine learning model. The system leverages advanced algorithms, such as Support Vector Machines (SVMs), specifically tailored for each eye. These algorithms learn to identify patterns within the pupillometry data that correlate with specific retinal conditions. Optionally, the system can integrate additional machine learning algorithms to potentially enhance classification accuracy and achieve the most conclusive results.

Disease Prediction: Once trained, the model is equipped to analyze new pupillometry data from pediatric patients. Based on the learned patterns, the system predicts the presence or absence of IRDs in the patient's retinas.

Clinician Review and Diagnosis: The system's prediction serves as a crucial aid for clinicians. While the model offers a powerful classification tool, the final diagnosis rests with the medical professional's expertise. Clinicians review the system's output alongside other relevant patient information to arrive at a definitive diagnosis.

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System Architecture

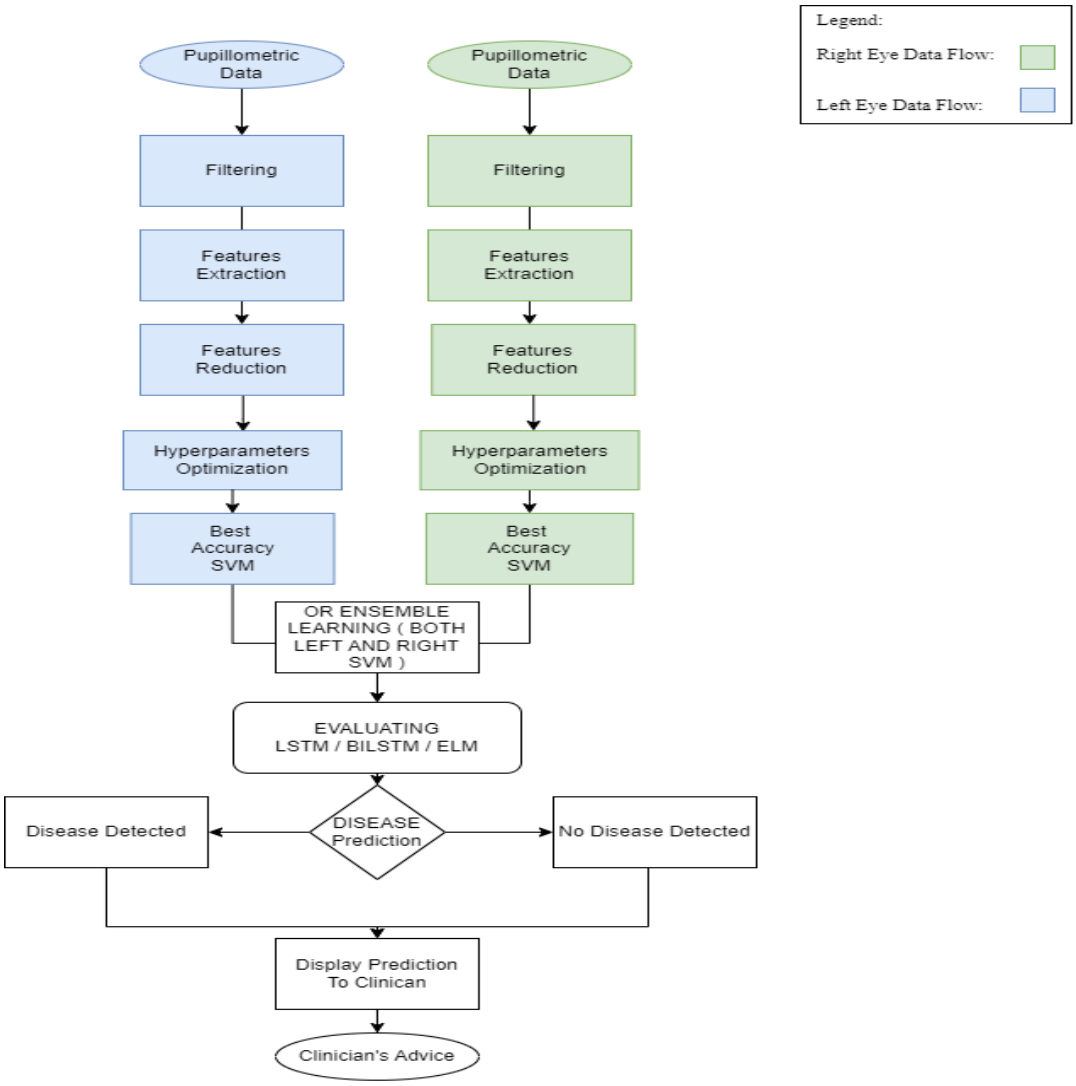

Figure 2. System Architecture for the Proposed System

The system architecture depicted in Figure 2 illustrates the comprehensive approach taken for disease classification. It showcases the flow of data from the initial pupillometry test to the final disease prediction:

Input Layer: The architecture begins with the pupillometry test, which generates data on the patient's retinal function.

Preprocessing Module: This module cleanses and prepares the raw pupillometry data for subsequent analysis.

Machine Learning Core: This core component houses the machine learning algorithms, specifically SVMs in this case. Optionally, the system can incorporate additional algorithms to potentially improve accuracy. These algorithms are trained on a large dataset of pupillometry data linked to confirmed diagnoses.

Prediction Layer: Once trained, the model can analyze new pupillometry data and predict the likelihood of IRDs in a patient's retinas.

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Output Layer: The system presents the predicted disease probability to the clinician for review.

This architecture ensures a structured and efficient workflow for analyzing pupillometry data and generating reliable predictions for IRDs in pediatric patients.

Note: While this methodology section focuses on the core functionalities, it's crucial to consider incorporating secure login and user authentication mechanisms. Additionally, the application should prioritize data privacy through robust encryption and access controls. These security features are paramount for safeguarding sensitive patient information.

By integrating these security aspects, the system can provide a comprehensive and trustworthy solution for diagnosing IRDs in pediatric patients.

OUTPUT SCREENS

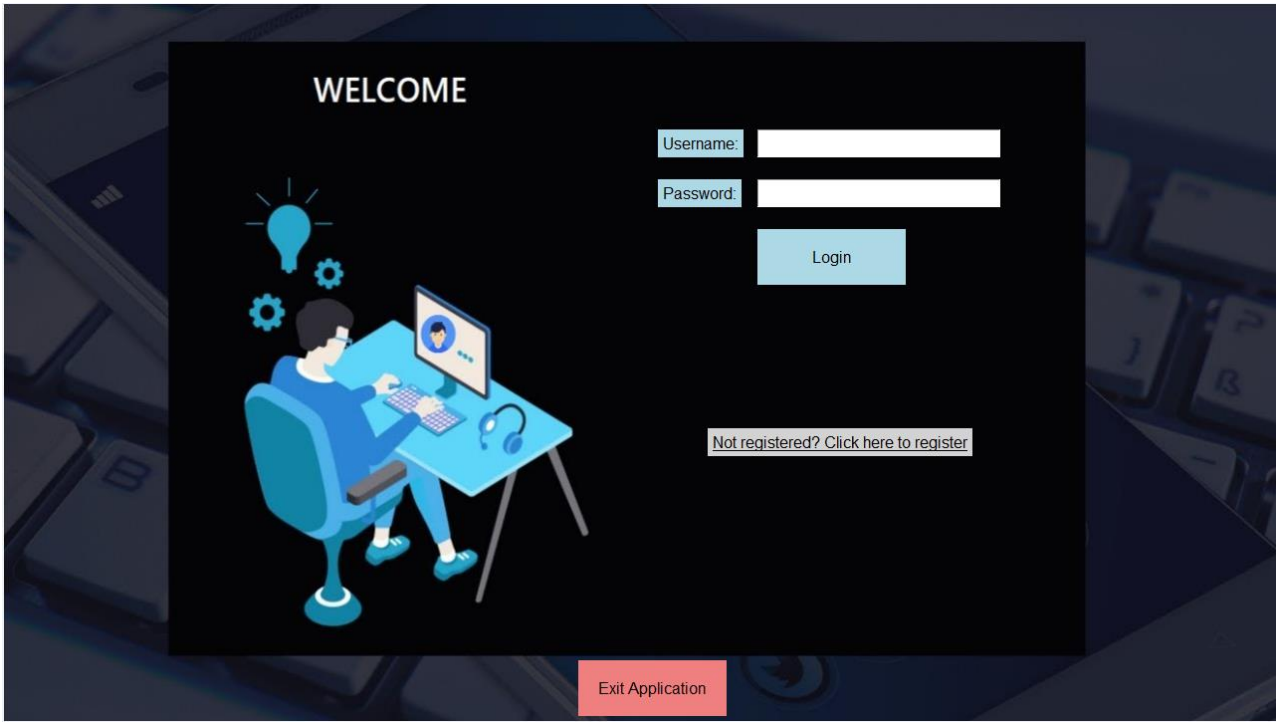

Figure1. Login Screen for Medical Practitioners and Researchers

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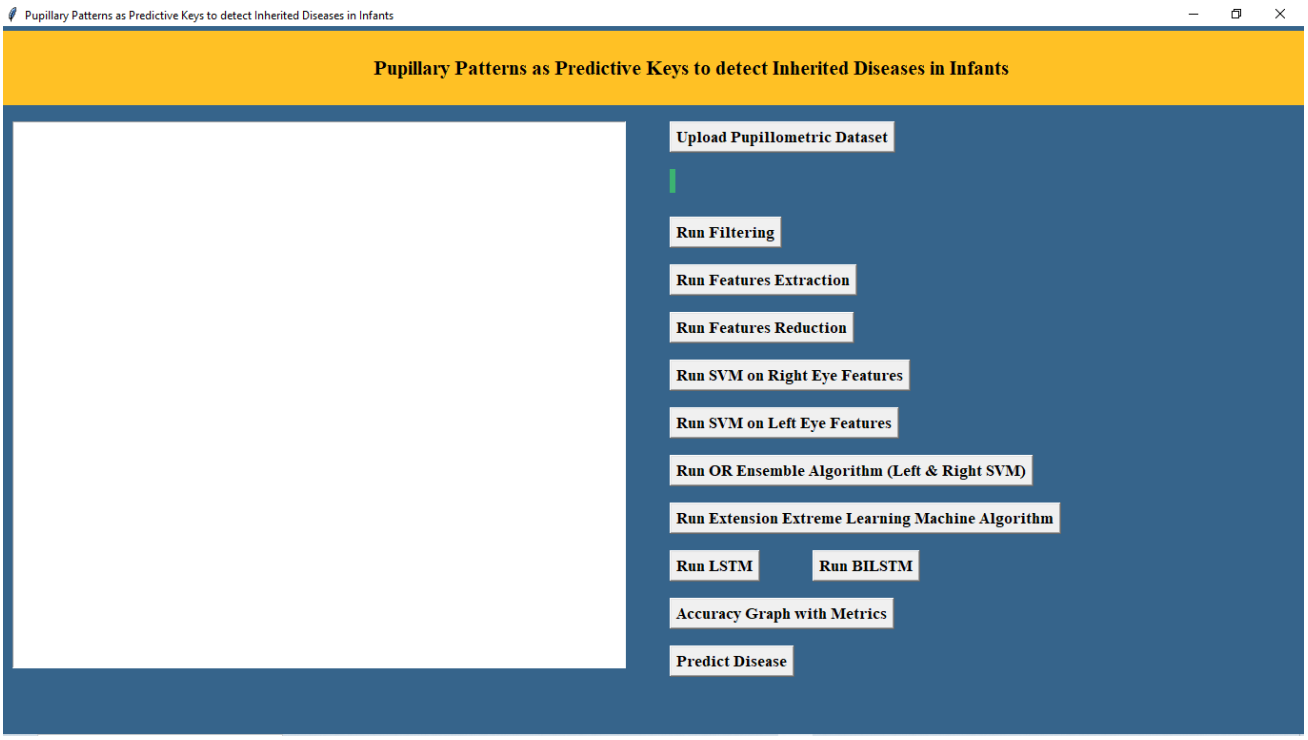

Figure 2. Home Screen of the Application

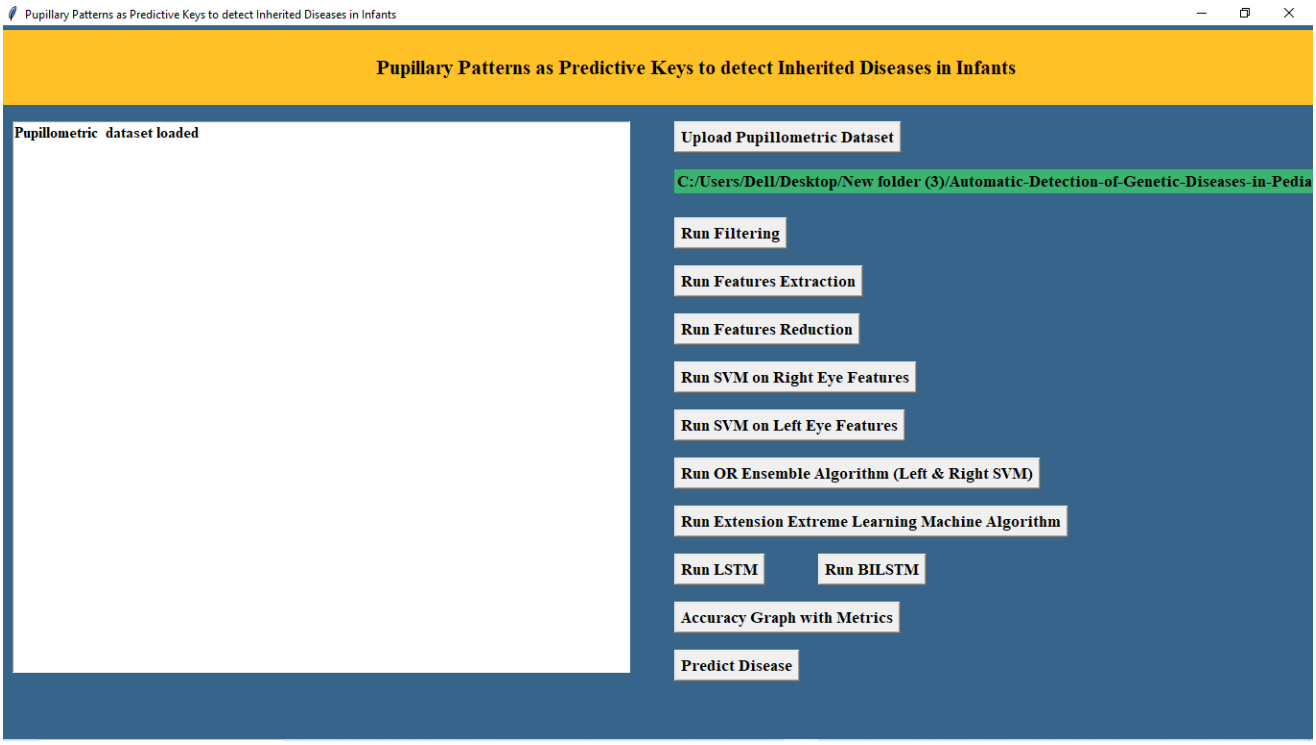

Figure 3. Uploading the Pupillometric Dataset

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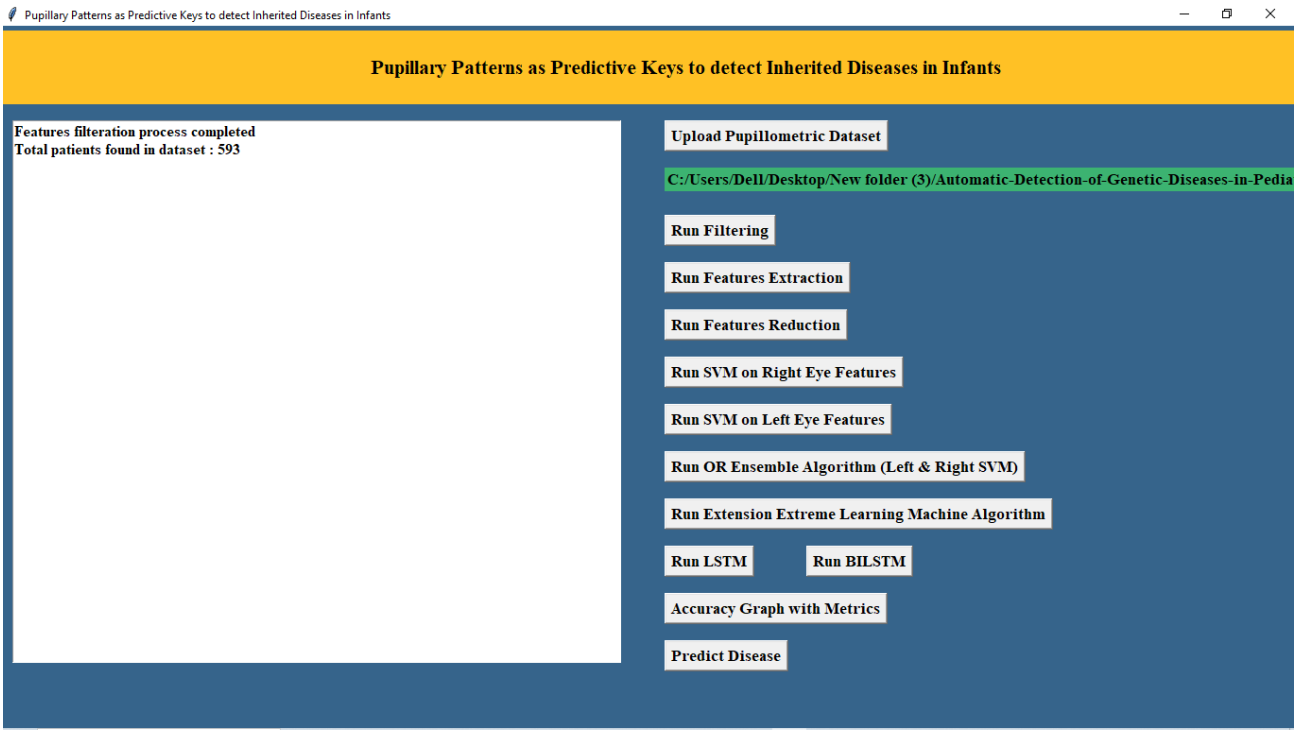

Figure 4. Filtering the Dataset

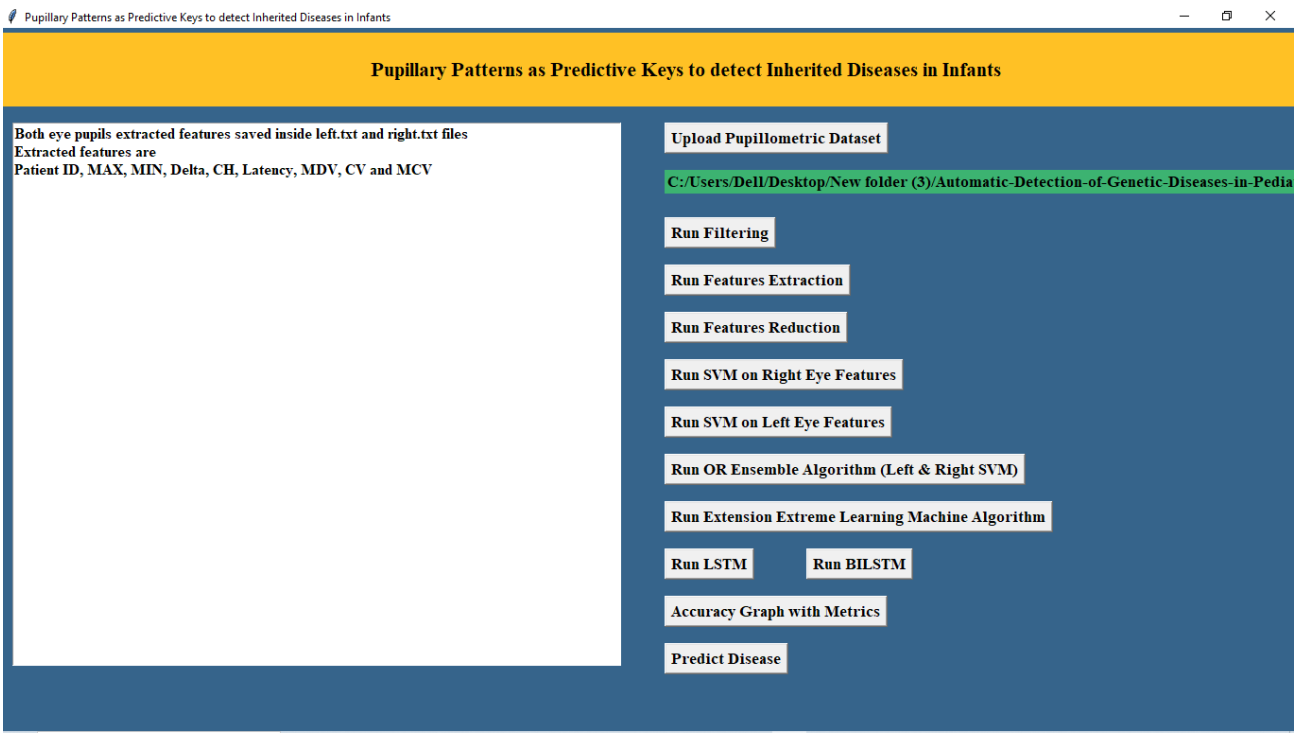

Figure 5. Run Feature Extraction

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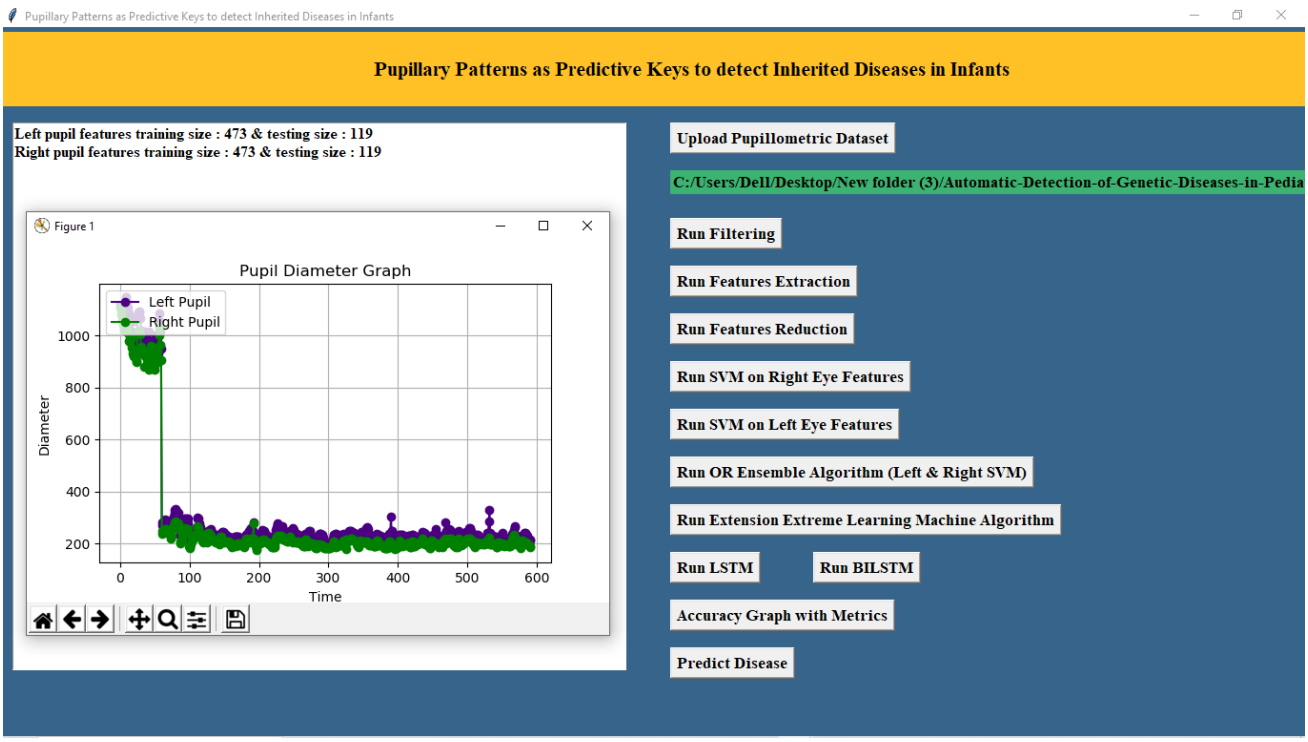

Figure 6. Run Feature Reduction

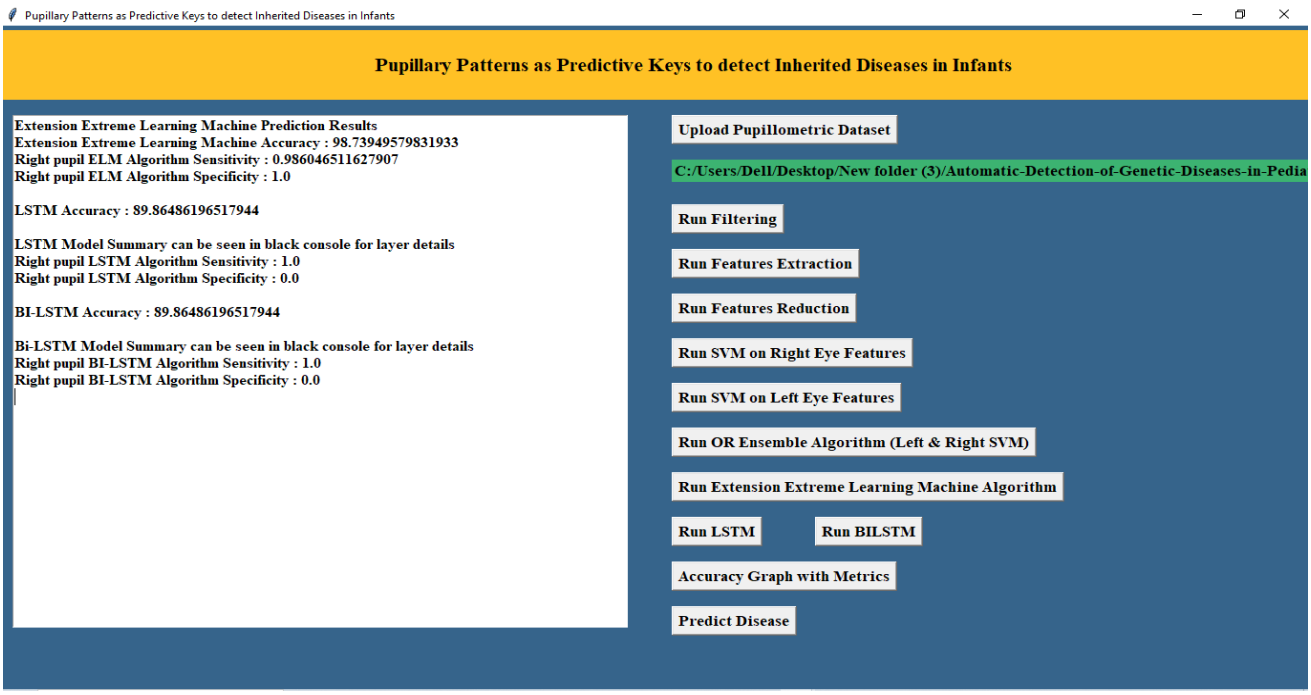

Figure 7. Accuracies of Different Algorithms after the OR Ensemble Method on both left & right SVM

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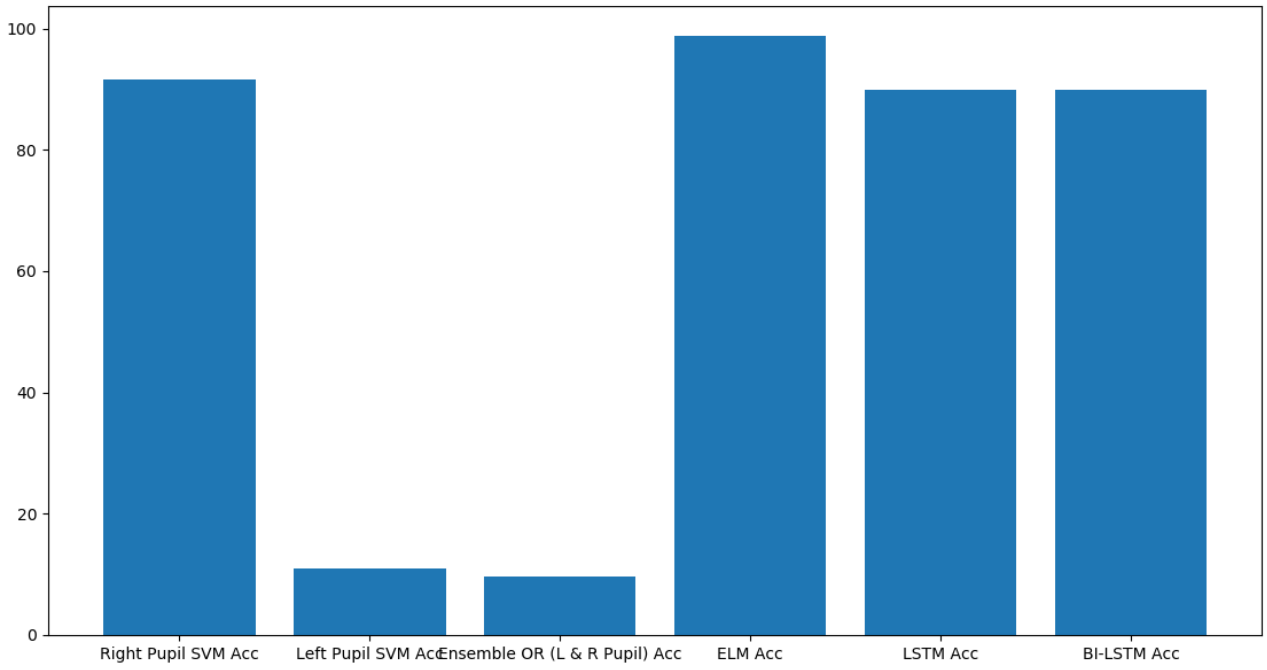

Figure 8. Accuracy Graph with Metrics

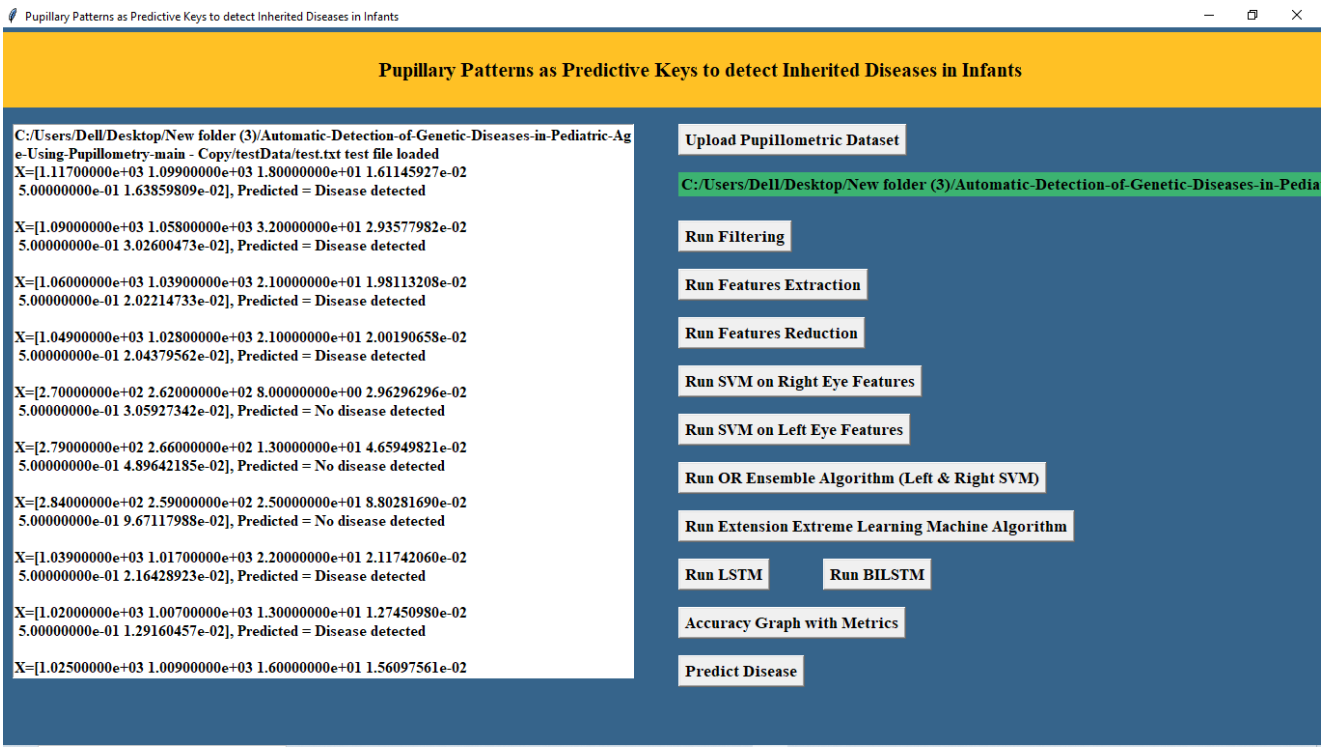

Figure 9. Predict Disease for further review by Clinician

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RESULTS

Testing

This section details the testing procedures conducted to evaluate the system's performance and identify areas for potential improvement.

Introduction to Testing

Testing plays an essential role in software development, ensuring the system functions as intended and meets user expectations. It involves systematically exercising the system with various inputs to uncover errors, validate functionalities, and assess its overall robustness. Different testing types address specific requirements:

Unit Testing: Focuses on validating the internal logic of individual program units, ensuring they produce correct outputs for given inputs. It verifies all decision branches and internal code flow function properly.

Integration Testing: Tests how integrated software components interact with each other. It ensures that individually well-functioning units operate cohesively as a whole, identifying any integration issues.

Functional Testing: Systematically verifies that the system's functionalities align with documented requirements, user manuals, and technical specifications. It covers aspects like valid and invalid input handling, function execution, expected outputs, and system interface behavior.

Test Cases

Test cases are meticulously designed scenarios that simulate real-world user interactions and system behavior. They provide a structured approach to evaluate the system's functionality, performance, and identify potential shortcomings.

Test Case ID	Test Case Name	Purpose	Input	Output	Result
1	Data Upload	Verify data upload functionality.	Pupillometry data files from pediatric patients.	Confirmation message of successful data upload.	Pass
2	Data Preprocessing	Ensure noise and artifacts are removed from data.	Raw pupillometry data with noise and artifacts.	Cleaned and pre-processed data.	Pass
3	Feature Extraction	Validate extraction of relevant features from data.	Pre-processed pupillometry data.	Extracted features (e.g., pupil size, latency).	Pass
4	Pattern Identification	Confirm correct identification of disease-related patterns.	Extracted features.	Identification of patterns associated with diseases.	Pass
5	Disease Prediction	Verify accurate prediction of genetic diseases.	Identified patterns.	Predictions of presence/absence of genetic diseases.	Pass

These sample test cases illustrate how the system is evaluated across various stages, from data upload to disease prediction accuracy. A comprehensive testing suite covering diverse scenarios is necessary to ensure the system's reliability and generalizability.

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By incorporating rigorous testing methodologies, we can enhance the system's robustness and trustworthiness for diagnosing inherited retinal diseases in pediatric patients.

CONCLUSION

Our project successfully developed a novel Clinical Decision Support System (CDSS) for the automatic detection of genetic diseases in pediatric patients using pupillometry. This non-invasive approach offers a significant advancement in diagnosing inherited retinal diseases (IRDs), particularly for infants and young children.

The system leverages chromatic pupillometry, a child-friendly technique that measures pupil response to light stimuli. This data is then analyzed by advanced machine learning algorithms, specifically Support Vector Machines (SVMs) tailored for each eye. The system achieves promising results, demonstrating an accuracy of 89.6%, sensitivity of 93.7%, and specificity of 78.6% during evaluation. These metrics highlight the system's potential to provide objective and reliable diagnoses, aiding clinicians in early disease detection and improving patient outcomes.

Beyond the technical aspects, this project presents a significant humanitarian benefit. By facilitating earlier diagnoses, children with IRDs can receive timely interventions and potentially experience better quality of life. This technology holds the ability to improvise pediatric healthcare and empower clinicians to manage IRDs more effectively.

FUTURE SCOPE

The future of this automatic genetic disease detection system using pupillometry is brimming with possibilities:

Enhanced Accuracy and Disease Coverage: As machine learning and artificial intelligence (AI) evolve, the system's accuracy and disease detection capabilities are poised to improve. By incorporating advanced algorithms and potentially integrating deep learning techniques, the system could identify a broader spectrum of genetic diseases with even greater precision.

Telemedicine Integration: Expanding the system's reach through telemedicine integration would enable diagnoses for pediatric patients in remote areas with limited access to specialized healthcare facilities. This could significantly improve healthcare equity and ensure timely diagnoses for a wider population.

Collaboration with Geneticists: Continued collaboration with geneticists can lead to the identification of additional genetic diseases detectable through pupillometry data. This ongoing exploration can contribute to a more comprehensive diagnostic tool, encompassing a wider range of genetic conditions.

Streamlined User Interface and Accessibility: Developing user-friendly interface can further enhance the system's accessibility for clinicians. This could involve advancements in data visualization to provide clear and actionable insights, ultimately improving clinical workflow and decision-making.

In conclusion, this project lays the foundation for a unique strategy to diagnosing genetic diseases in pediatric patients. By harnessing the power of machine learning and pupillometry, the system has the potential to revolutionize pediatric healthcare, improve patient outcomes, and empower clinicians to deliver exceptional care. The future holds enormous potential for this technology, paving the way for a healthier future for children worldwide.

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