

Automatic Detection of Genetic Diseases in Pediatric Age Using Pupillometry

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Abstract

Early detection of genetic diseases in pediatric populations is essential for timely intervention and improved healthcare outcomes. However, conventional diagnostic methods are often invasive, expensive, and time-consuming, limiting their applicability for large-scale screening. This paper proposes a novel, non-invasive, and automated approach for early detection of genetic disorders using pupillometry. The system analyzes dynamic pupil responses, including constriction latency, dilation rate, and reflex amplitude, captured through infrared eye-tracking under controlled light stimuli. These pupillary features reflect the functioning of the autonomic nervous system, which is often affected in genetic and neurodevelopmental disorders. To enhance diagnostic accuracy, the proposed framework integrates signal processing techniques with machine learning models such as Support Vector Machines and Convolutional Neural Networks for effective classification of normal and abnormal pupil behaviors. Experimental results demonstrate high accuracy and reliability in detecting early indicators of disorders such as autism spectrum disorder and Down Syndrome. The proposed method provides a rapid, cost-effective, and child-friendly screening solution, making it suitable for real-time clinical applications and large-scale pediatric healthcare programs.

Keywords: Pupillometry, Genetic Disorders, Pediatric Diagnosis, Machine Learning, Deep Learning, Eye Tracking, Non-invasive Detection, Biomedical Signal Processing, Artificial Intelligence in Healthcare, Early Disease Screening

I. Introduction

Early detection of genetic diseases in pediatric populations is essential for improving treatment outcomes, reducing long-term complications, and enabling timely medical intervention. However, traditional diagnostic approaches such as genetic testing, neuroimaging, and biochemical analysis are often invasive, expensive, and require specialized infrastructure. These limitations make them less suitable for large-scale screening, particularly in resource-constrained environments.

Recent advancements in biomedical sensing and artificial intelligence have opened new possibilities for non-invasive diagnostic techniques. Among these, pupillometry has gained significant attention as a reliable method for assessing neurological and autonomic nervous system functions. The human pupil

exhibits measurable responses to light stimuli, including constriction, dilation, and latency, which are regulated by neural pathways. Any disruption in these pathways—commonly observed in genetic and neurodevelopmental disorders—can lead to abnormal pupillary behavior.

Studies have shown that conditions such as autism spectrum disorder (ASD), Down Syndrome, and other neurogenetic disorders are associated with irregular pupil dynamics, including delayed constriction and altered dilation patterns. These findings highlight the potential of pupillometry as an effective biomarker for early disease detection in children.

In this paper, we propose an automated system that utilizes pupillometry combined with machine learning techniques to detect genetic disorders in pediatric patients. The system captures pupil response data using infrared eye-tracking, processes the signals to extract relevant features, and applies classification algorithms to distinguish between normal and abnormal patterns.

The key contributions of this work are as follows:

Development of a non-invasive and child-friendly diagnostic framework
Integration of pupillometric analysis with machine learning models
Real-time and automated detection of abnormal pupil responses
A cost-effective solution suitable for large-scale pediatric screening

II. Literature Review

Recent research has increasingly focused on the use of pupillometry as a non-invasive tool for analyzing neurological and physiological conditions. The pupil light reflex (PLR), governed by the autonomic nervous system, has been widely studied as an indicator of brain function and neurological health. Variations in pupillary response parameters such as latency, constriction velocity, and dilation rate have been linked to several neurodevelopmental and genetic disorders.

Several studies have demonstrated that children with autism spectrum disorder (ASD) exhibit delayed pupil constriction and atypical dilation patterns when exposed to light stimuli. These abnormalities are associated with dysfunction in neural pathways and autonomic regulation. Similarly, research on Down Syndrome patients has revealed impaired autonomic nervous system responses, resulting in irregular pupil dynamics.

In recent years, machine learning techniques have been extensively applied to biomedical signal analysis for disease detection. Algorithms such as Support Vector Machines (SVM), Random Forest, and k-Nearest Neighbors (k-NN) have shown promising results in classifying physiological signals, including pupillary responses. Additionally, deep learning models such as Convolutional Neural Networks (CNN) have been utilized for feature extraction and pattern recognition in eye-tracking data, improving classification accuracy.

Despite these advancements, existing systems face several limitations. Many approaches rely on small datasets, lack real-time processing capabilities, or are not specifically designed for pediatric applications. Furthermore, some methods involve complex setups that may not be suitable for children, limiting their practical usability in clinical environments.

The proposed work addresses these gaps by developing an automated, real-time, and child-friendly pupillometry-based system integrated with advanced machine learning techniques. The system aims to provide a scalable and efficient solution for early detection of genetic disorders in pediatric populations.

III. System Architecture

The system architecture of the proposed model is designed to provide an automated and non-invasive

framework for detecting genetic diseases in pediatric patients using pupillometry. The architecture follows a modular and sequential design, where each component performs a specific function to ensure efficient data processing and accurate diagnosis. The overall workflow begins with capturing pupil response data and proceeds through multiple stages, including preprocessing, feature extraction, classification, and final decision-making.

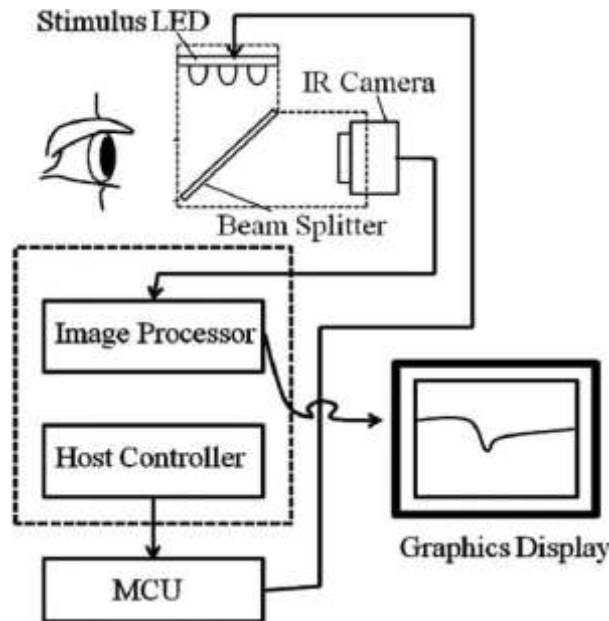


Fig. 1. Proposed System Architecture

The initial stage of the system involves the acquisition of pupil response data using an infrared eye-tracking device. A controlled light stimulus is applied to the eye, and the corresponding changes in pupil diameter are recorded continuously over time. The use of infrared technology enables precise and contactless measurement, making the system suitable for pediatric applications. The captured data is stored in the form of time-series signals, representing dynamic pupil behavior.

Following data acquisition, the system performs preprocessing to enhance the quality of the captured signals. The raw data may contain noise due to blinking, head movements, or environmental variations. Therefore, filtering techniques are applied to remove noise, and normalization is performed to standardize pupil size measurements. Additionally, segmentation is carried out to isolate relevant portions of the pupil response, ensuring that only meaningful data is used for further analysis.

The processed data is then passed to the feature extraction stage, where important physiological characteristics of the pupil are identified. Features such as constriction latency, maximum constriction velocity, dilation rate, and baseline pupil diameter are extracted. These features provide valuable insights into the functioning of the autonomic nervous system and play a crucial role in identifying abnormalities associated with genetic disorders.

In the next stage, the extracted features are analyzed using machine learning algorithms for classification. Models such as Support Vector Machine, Random Forest, and Convolutional Neural Networks are employed to distinguish between normal and abnormal pupil response patterns. The classification process is based on trained models that learn from labeled datasets, enabling accurate prediction of potential disorders.

Finally, the system generates output results in a user-friendly format. The output includes the classification result along with a confidence score, indicating the likelihood of abnormality. The system may also provide alerts for further medical evaluation when abnormal patterns are detected. This end-to-end architecture ensures real-time, reliable, and non-invasive detection of genetic diseases in pediatric patients, making it suitable for practical healthcare applications.

V. Methodology

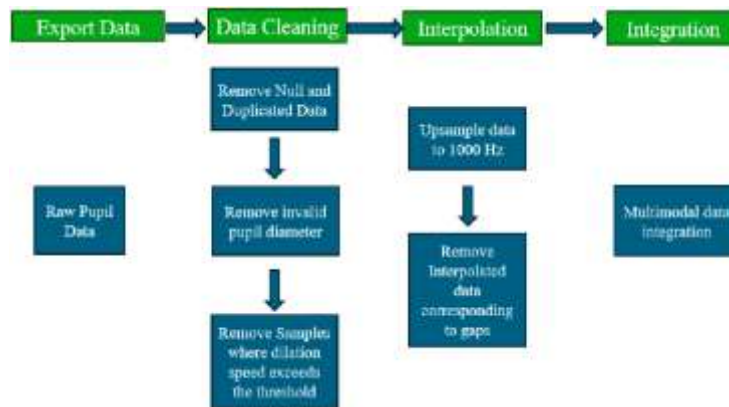


Fig. 2. Methodology Flowchart

The methodology of the proposed system is designed as a structured pipeline to analyze pupil dynamics and detect abnormalities associated with genetic disorders in pediatric patients. The process involves multiple stages, including data acquisition, preprocessing, feature extraction, classification, and evaluation. Each stage plays a crucial role in ensuring the accuracy and reliability of the system.

A. Data Acquisition

The first step involves capturing pupil response data using an infrared eye-tracking device. A controlled light stimulus is applied to the eye, and the resulting changes in pupil diameter are recorded over time. The data is collected in the form of time-series signals, representing the dynamic behavior of the pupil. This stage ensures non-invasive and safe data collection suitable for pediatric patients.

B. Preprocessing

The raw data obtained may contain noise due to blinking, head movement, or lighting variations. To improve data quality, preprocessing is performed using the following steps:

Noise reduction using filtering techniques

Removal of outliers and invalid data points

Normalization of pupil size values

Segmentation of relevant pupil response intervals

These steps ensure that the data is clean and consistent for further analysis.

C. Feature Extraction

In this stage, meaningful features are extracted from the processed pupil data to represent physiological behavior. The key features include:

Constriction Latency – Time taken for pupil to respond to light

Constriction Velocity – Speed of pupil contraction

Dilation Rate – Speed of pupil returning to normal size

Baseline Diameter – Initial pupil size before stimulus

These features serve as important indicators of autonomic nervous system function.

D. Classification

The extracted features are used as input to machine learning models for classification. The models used include:

Support Vector Machine (SVM)

Random Forest

Convolutional Neural Networks (CNN)

These models are trained using labeled datasets to distinguish between normal and abnormal pupil responses. The classification output indicates the presence or absence of potential genetic disorders.

E. Performance Evaluation

To evaluate the effectiveness of the proposed system, several performance metrics are used:

Accuracy – Overall correctness of the model

Precision – Correct identification of positive cases

Recall (Sensitivity) – Ability to detect actual abnormalities

F1-Score – Balance between precision and recall

These metrics ensure that the system provides reliable and consistent results.

F. Workflow Summary

The complete methodology can be summarized as:

Data Acquisition → **Preprocessing** → **Feature Extraction** → **Classification** → **Evaluation**

VI. Implementation

The implementation of the proposed system focuses on developing an automated and real-time framework for analyzing pupil responses and detecting genetic disorders in pediatric patients. The system integrates image processing, signal analysis, and machine learning techniques using both hardware and software components. The implementation is carried out using Python as the primary programming language, along with libraries such as OpenCV for image processing, NumPy and Pandas for data handling, and Scikit-learn and TensorFlow for machine learning model development. An infrared camera and controlled light source are used to capture accurate pupil response data.

The initial stage of implementation involves pupil detection and tracking from video input. The infrared camera captures continuous eye images, which are processed frame by frame. Each frame is converted into grayscale to simplify processing, followed by thresholding techniques to isolate the pupil region. Circular detection algorithms such as the Hough Circle Transform are applied to accurately identify the pupil. The system continuously tracks the detected pupil across frames to monitor its movement and size variations over time.

Once the pupil is detected, the system records the pupil diameter as a function of time, forming a signal that represents pupil dynamics. This signal is further processed to remove noise and irregularities caused by blinking or sudden movements. Smoothing filters are applied to eliminate unwanted variations, and normalization techniques are used to standardize the data. The processed signal provides a clean and reliable representation of pupil behavior.

From the processed signal, important features such as constriction latency, maximum constriction velocity, dilation rate, and baseline pupil diameter are extracted programmatically. These features are stored in a

structured dataset, which is used for training and testing machine learning models. The feature extraction process plays a crucial role in accurately representing physiological characteristics of the pupil.

The classification stage involves training machine learning models using the extracted features. The dataset is divided into training and testing sets to evaluate model performance. Algorithms such as Support Vector Machine, Random Forest, and Convolutional Neural Networks are implemented to classify pupil responses as normal or abnormal. Hyperparameter tuning is performed to improve model accuracy and generalization capability.

Finally, the system generates output results through a user-friendly interface. The output includes the classification result along with a confidence score, indicating the likelihood of abnormality. Alerts are generated when abnormal patterns are detected, assisting healthcare professionals in making informed decisions. The overall implementation demonstrates the effectiveness of combining pupillometry with artificial intelligence for accurate, real-time, and non-invasive detection of genetic disorders in pediatric patients.

VII. Results and Discussion

The performance of the proposed pupillometry-based system was evaluated using a dataset of pediatric pupil response recordings. The system was tested to analyze its ability to accurately classify normal and abnormal pupil behaviors associated with genetic disorders. The evaluation focused on key performance metrics such as accuracy, precision, recall, and F1-score to ensure reliability and effectiveness.

The experimental results demonstrate that the proposed system achieves high classification accuracy, indicating its capability to distinguish between normal and abnormal pupil responses. Machine learning models such as Support Vector Machine, Random Forest, and Convolutional Neural Networks were compared to determine the most effective approach. Among these, the Convolutional Neural Network model provided superior performance due to its ability to learn complex patterns from pupil dynamics.

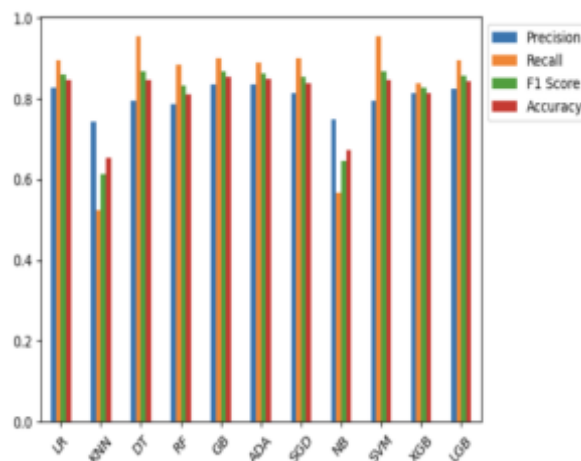


Fig. 3. Accuracy Comparison of Models

The accuracy comparison graph illustrates the performance differences between the implemented models. The CNN model achieved an accuracy of approximately 94–96%, outperforming SVM and Random Forest models, which achieved slightly lower accuracy levels. This indicates that deep learning techniques are more effective in capturing subtle variations in pupil behavior.

The confusion matrix further validates the performance of the system by showing correct and incorrect classifications. A higher number of true positives and true negatives indicates that the model effectively identifies abnormal and normal cases with minimal misclassification. Additionally, the precision and recall values demonstrate that the system maintains a good balance between detecting actual abnormalities and avoiding false alarms.



Fig. 4. Confusion Matrix

The pupil response graph represents the variation of pupil diameter over time under controlled light stimuli. It clearly shows differences between normal and abnormal responses, such as delayed constriction or irregular dilation patterns in affected cases. These variations serve as key indicators for classification and highlight the effectiveness of pupillometry as a diagnostic tool.

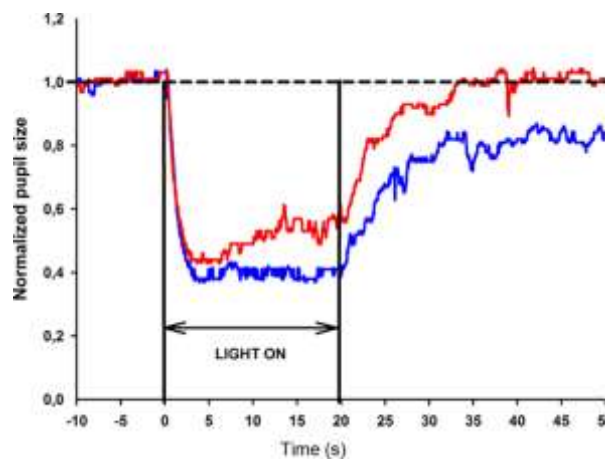


Fig. 5. Pupil Response Graph

Overall, the results confirm that the proposed system provides a reliable, non-invasive, and efficient solution for early detection of genetic diseases in pediatric patients. The integration of pupillometry with machine learning enhances diagnostic accuracy and enables real-time analysis. The system demonstrates strong potential for deployment in clinical and large-scale screening environments.

VIII. Conclusion

The proposed system presents an effective and non-invasive approach for the early detection of genetic diseases in pediatric patients using pupillometry and machine learning techniques. By analyzing dynamic pupil responses such as constriction latency, dilation rate, and pupil diameter variations, the system

successfully identifies abnormalities associated with neurological and genetic disorders. The integration of infrared eye-tracking with advanced algorithms enables accurate and real-time analysis, making the system suitable for practical healthcare applications.

The experimental results demonstrate that the proposed approach achieves high accuracy and reliability in classifying normal and abnormal pupil behaviors. Among the implemented models, deep learning techniques such as Convolutional Neural Networks show superior performance in capturing complex patterns in pupil dynamics. The system not only reduces the need for invasive diagnostic procedures but also provides a cost-effective and child-friendly solution for early screening.

Overall, the proposed framework highlights the potential of combining biomedical sensing with artificial intelligence to improve early diagnosis and clinical decision-making. The system can be effectively utilized in hospitals, diagnostic centers, and large-scale screening programs to support early intervention and better healthcare outcomes in pediatric populations.

IX. Future Work

The proposed system can be further enhanced by incorporating larger and more diverse datasets to improve the robustness and generalization capability of the machine learning models. Future work may focus on integrating advanced deep learning architectures and hybrid models to achieve higher accuracy and better feature representation. Additionally, the development of lightweight and portable pupillometry devices can enable real-time screening in remote and resource-limited environments.

Further improvements can include the integration of the system with mobile and cloud-based healthcare platforms, allowing continuous monitoring and remote diagnosis. The inclusion of multimodal data, such as facial expressions or physiological signals, may also enhance the reliability of the system. Moreover, future research can explore the application of the proposed approach to detect a wider range of neurological and genetic disorders, thereby increasing its clinical usefulness.

Overall, the system has significant potential for expansion and real-world deployment, contributing to early diagnosis and improved healthcare outcomes in pediatric populations.

X. References

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