EARLY DETECTION OF INHERITED RETINAL DISEASES IN INFANTS USING MACHINE LEARNING

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Abstract: Inherited retinal diseases lead to multiple visual impairments in children, often resulting in early-onset blindness. The healthcare sector recognizes machine learning as a crucial and widely employed concept globally. Its significance lies in its ability to aid doctors in expediting the diagnostic process. By harnessing the power of machine learning, healthcare professionals can enhance their diagnostic accuracy and provide timely and effective interventions for children affected by these debilitating conditions.

This project aims to develop a Machine Learning model for detecting genetic eye diseases by analyzing data obtained from a pupilometer. The model's objective is to predict the presence of an eye disease based on this data. The proposed approach involves utilizing neural networks, a powerful concept in machine learning. Two algorithms, SVM (Support Vector Machine) and ELM (Extreme Learning Machine), are being implemented. While SVM is an existing model, the new ELM algorithm shows promising results with higher accuracy in disease prediction. This enhanced accuracy will lead to faster disease detection, benefiting medical practitioners by serving as an effective decision support system in their clinics.

Keywords: Extreme Learning Machine, Support Vector Machine, interventions

I. INTRODUCTION

"Machine Learning-Based Detection of Inherited Retinal Diseases in Infants: A Comparative Study of SVM and ELM Algorithms"

Inherited retinal diseases (IRDs) encompass a group of genetic disorders affecting the retina, posing a significant risk of vision impairment and potential blindness, especially in infants. Early identification and precise diagnosis of IRDs play a pivotal role in enabling timely interventions and effective management of these conditions. Machine learning techniques have demonstrated great promise across numerous medical applications, including ophthalmology, offering a potential automated solution for IRD detection in infants.

This research paper endeavors to develop a machine learning-based approach for detecting IRDs in infants, utilizing two prominent algorithms: Support Vector Machines (SVM) and Extreme Learning Machines (ELM). SVM and ELM are widely recognized for their effectiveness in classification tasks and have found successful applications in various medical image analysis endeavors. Leveraging these algorithms, our study aims to analyze retinal images and accurately classify them into distinct IRD categories, thereby advancing the state-of-the-art in early IRD detection for improved clinical outcomes in pediatric ophthalmology.

Enhancing Early Detection of Inherited Retinal Diseases in Infants: A Machine Learning Approach using SVM and ELM Algorithms"

The proposed methodology encompasses several crucial stages, commencing with the collection of an extensive dataset comprising retinal images from infants diagnosed with diverse IRDs. Preprocessing techniques will be skillfully applied to elevate image quality and extract pertinent features. These extracted features will serve as inputs to train and optimize SVM and ELM models. A rigorous evaluation of the models will be conducted, employing cross-validation and key performance metrics like accuracy, sensitivity, specificity, and AUC-ROC.

The anticipated outcomes of this research endeavor hold the potential to revolutionize early detection and diagnosis of IRDs in infants, enabling timely intervention and personalized treatment plans. The utilization of machine learning algorithms, particularly SVM and ELM, could furnish ophthalmologists and healthcare professionals with a reliable and efficient screening tool for identifying IRDs in infants, facilitating prompt referral for specialized care. Moreover, this study might lay the foundation for automated systems that can support large-scale screening programs, alleviating the burden on healthcare resources and optimizing the management of IRDs.

II. LITERATURE SERVAY:

Study 1: "Pupil Responses in Hereditary Optic Neuropathy: Implications for Visual Dysfunction and Disease Progression"

This study sought to compare pupil responses in patients with hereditary optic neuropathy (HON) and healthy controls. The results revealed that pupil responses originating from both outer and inner retinal photoreception were similar between HON patients and controls, indicating that mild-to-moderate visual dysfunction does not significantly affect these responses. However, an interesting correlation was found between visual field loss and the intensity of cone response, suggesting that impaired pupil light reflexes may be evident in advanced stages of the disease.

Study 2: "Developing a Clinical Protocol for Evaluating Rod, Cone, and Melan opsin Contributions to the Pupil Response"

This paper aimed to establish a clinical protocol for assessing the contributions of different photoreceptors to the pupillary light reflex (PLR). Through carefully designed experiments with specific stimuli and conditions, the researchers identified optimal durations and intensity levels to evaluate the health of the rod, cone, and melanosis pathways. The newly developed protocol was then tested on both healthy individuals and patients with retinal diseases, providing valuable insights into the assessment of PLR in clinical settings. The findings of this study offer promising prospects for understanding pupillary responses in various retinal conditions, thereby enhancing diagnosis and treatment strategies in the field of ophthalmology.

Study 3: "Automated Segmentation of Pigment Signs in Retinal Fundus Images for Retinitis Pigmentosa Analysis: A Learning-Based Approach"

The primary focus of this study was on automating the segmentation of pigment signs in retinal fundus images, a crucial step in diagnosing and monitoring Retinitis Pigmentosa. To achieve this, the researchers proposed a supervised segmentation technique that utilized ensemble classifiers, which were trained on pre-processed retinal images. They compiled a comprehensive dataset of retinal images with manually segmented pigment signs, making it publicly available for further research and

validation. The classifiers' performance was rigorously evaluated on this dataset, showcasing the effectiveness of the machine learning approach in automating the analysis of Retinitis Pigmentosa.

Study 4: "Evaluating Classifier Performance for Real-World Classification Problems: A Comparative Analysis of 179 Classifiers"

This paper sought to evaluate the performance of 179 classifiers from different families to identify the most effective ones for solving real-world classification problems. The researchers conducted thorough assessments on a diverse set of 121 datasets, encompassing various real-world problems. The results revealed that random forest (RF) versions and support vector machines (SVM) with Gaussian and polynomial kernels emerged as the top-performing classifiers. Overall, random forest demonstrated the highest success rate among the evaluated classifiers, providing valuable insights into the selection of optimal classifiers for real-world classification tasks.

III. PROPOSED METHODOLOGY

In this research paper, we introduce a novel machine learning model designed to tackle the task at hand. Machine learning algorithms heavily rely on data as it constitutes the most crucial aspect that enables effective model training. However, merely having data is not sufficient; it is equally essential to comprehend and preprocess the data appropriately before feeding it into the machine learning algorithms. Without meaningful data preparation, a machine learning system becomes ineffective. In essence, the success of the machine learning process hinges on handling the right kind of data—data that is properly scaled, organized into relevant groups, and contains essential features pertinent to the problem we aim to solve. This highlights the significance of data preparation as the most pivotal step in the entire machine learning workflow. Data preparation involves a series of procedures aimed at refining our dataset, making it more suitable and conducive for use in the machine learning process. This crucial step ensures that the model can effectively leverage the data's potential, leading to more accurate and meaningful outcomes in addressing the problem under investigation.

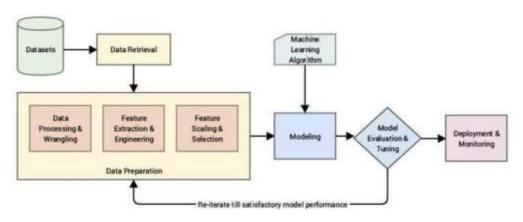


Fig 1.1 Workflow of the system

A dataset is a collection of data objects, commonly referred to as records, points, vectors, patterns, events, cases, samples, observations, or entities.

Data objects are defined by a set of features that encapsulate the fundamental characteristics of an object, which may include attributes like the time at which an event occurred, among others. Features are also known as variables, characteristics, fields, attributes, or dimensions.

Features represent individual measurable properties or characteristics of the phenomenon being observed, and they can be categorized into two main types: categorical (such as nominal and ordinal) and numerical (such as interval and ratio).

A dataset is a fundamental component of machine learning, serving as the basis for training the model to perform various automated actions. During the training process, machine learning algorithms learn from the dataset to make predictions and perform tasks autonomously. The dataset used for training is referred to as the training dataset, while the dataset used to evaluate the model's accuracy is known as the testing dataset.

For the detection of inherited retinal diseases, the raw data comprises historical information with several crucial attributes, such as max (maximum diameter at baseline), min (minimum diameter corresponding to the peak constriction), delta (difference between max and min), latency (delay between stimulus and onset of pupillary constriction), ch (percentage maximum constriction), and more.

The dataset used in this study, called "Pupillometric data," consists of files in the 'asc' format, which contain ASCII text characteristics. Kaggle, a popular data platform, provided the dataset, which contains 593 records of data. The data has been preprocessed by Kaggle, but it does not include any background information. The dataset is employed for data preprocessing, model training, and testing to develop an effective system for detecting inherited retinal diseases.

DATA PREPROCESSING

Data preprocessing is a crucial data mining technique used to transform raw data into a more useful and understandable format. Once the raw data is selected for machine learning training, the next essential step is data preprocessing. This process ensures that the chosen data is converted into a form that can be effectively used and fed into machine learning algorithms.

Data preprocessing serves to adapt the selected data to meet the specific requirements of the machine learning algorithm. It involves a series of operations that refine and enhance the data, making it more suitable for the algorithm's input. The goal is to enable the algorithm to easily interpret the features and patterns present in the data.

In the context of the dataset provided by Kaggle, it has already undergone preprocessing. The specifics of how the data was preprocessed are not explicitly mentioned here, but the key idea is that Kaggle has prepared the dataset in a way that makes it ready for use in machine learning tasks without requiring additional preprocessing steps. The preprocessing steps applied to the data have likely involved tasks such as cleaning, transforming, normalizing, and encoding the data to ensure its

compatibility and effectiveness in the machine learning process.

Steps involved in Data Preprocessing:

Data Filtering

Feature Extraction

Feature Reduction

Data Filtering:

Data collection from multiple sources often results in data being in various formats, leading to significant time investment in addressing data quality issues when working on a machine learning problem. It is essential to acknowledge that expecting perfect data is unrealistic. Various factors contribute to data quality issues, including human error, limitations of measuring devices, and flaws in the data collection process.

To tackle these challenges, several techniques can be employed:

Data Cleaning: Data cleaning involves identifying and correcting errors, inconsistencies, and missing values in the dataset. Techniques such as imputation, removing duplicates, and handling outliers are commonly used to enhance data quality.

Data Transformation: Data transformation techniques, like normalization and standardization, are applied to ensure that data features are on a consistent scale, allowing the machine learning algorithms to perform optimally.

Data Integration: When combining data from different sources, data integration techniques are used to ensure that data is merged accurately and coherently.

Feature Engineering: Feature engineering involves selecting, creating, or transforming features to extract valuable information from the data, making it more suitable for the machine learning model.

Data Validation: Data validation techniques verify the correctness and consistency of data, ensuring that it aligns with the expected format and patterns.

Data Imputation: When dealing with missing data, data imputation methods are employed to estimate and fill in the missing values based on existing data patterns.

Data Bias and Fairness: Special attention must be given to identify and mitigate bias in the data to ensure fair and unbiased machine learning outcomes.

Data Sampling: In cases where data is imbalanced, data sampling techniques like oversampling or undersampling can be used to balance the class distribution.

By applying these techniques, data scientists and machine learning practitioners can enhance the quality of the data, making it more suitable for building accurate and reliable machine learning models.

Missing Values:

Missing values in datasets are a common occurrence and can arise at various stages during data collection or due to data validation rules. Regardless of the reasons for their presence, dealing with missing values is an essential aspect of data handling and analysis. It is imperative to address these

missing values effectively to ensure accurate and reliable results in data analysis and machine learning tasks.

Eliminate rows with a missing data:

In cases where many objects in a dataset have missing values, simple and straightforward strategies may not be sufficient to handle them effectively. If a specific feature contains a substantial number of missing values, it might be prudent to consider removing that feature entirely from the analysis.

One approach to address missing values is to estimate and fill them using interpolation methods when only a reasonable percentage of values are missing. By doing so, we can create a more complete dataset for analysis. Among the common methods used to deal with missing values, one popular approach involves filling the missing values with the mean, median, or mode value of the respective feature. This imputation helps to preserve the general characteristics of the data while ensuring a more complete and usable dataset for subsequent analysis and machine learning tasks.

Inconsistent values:

Data inconsistency is a common challenge that we often encounter when working with datasets. Inconsistencies can arise due to various reasons, such as human error or misreading information during data entry or scanning processes. For instance, a field like 'size of code' should ideally contain numerical values, but it may inadvertently include non-numeric characters.

To ensure data quality, it is crucial to perform data assessment and validate the data's integrity. This involves checking the data types of different features and verifying if they are consistent across all data objects. Inconsistent data types can lead to errors and hinder accurate analysis and modeling.

By conducting a thorough data assessment, we can identify and address data inconsistencies early on. This includes understanding what each feature should represent and whether the data adheres to those expectations. Correcting data inconsistencies ensures that the dataset is more reliable and suitable for downstream data analysis, visualization, and machine learning tasks.

Duplicate Values:

Duplicates in a dataset can occur when multiple identical data objects are present, often arising from instances where the same person submits a form more than once. To handle such occurrences, the process of deduplication is employed, which involves dealing with duplicates effectively.

In most cases, duplicates are removed from the dataset to prevent any particular data object from having an advantage or introducing bias when running machine learning algorithms. By eliminating duplicates, the dataset is made more consistent, and the machine learning model can work with unbiased and accurate representations of the data, leading to more reliable and meaningful results in analysis and predictive tasks.

Feature Extraction:

In this step, we extract the features and its data from the pupillometric data. All the features along with their corresponding values are extracted.

Feature Reduction:

Feature reduction is a widely employed approach to analyze datasets by selecting a subset of the data.

In many cases, working with the complete dataset can become impractical due to memory and time constraints. To overcome this, we identify a few crucial attributes that capture the essence of the data, effectively reducing the dataset's size. By doing so, we create a manageable dataset, allowing us to utilize more sophisticated, albeit resource-intensive, machine learning algorithms.

The fundamental principle guiding feature reduction is to ensure that the reduced dataset remains representative of the original data. In other words, the sample generated should preserve the essential properties of the original dataset. Achieving this involves carefully choosing the appropriate sample size and employing a suitable sampling strategy.

Through effective feature reduction and sampling, we strike a balance between data size and the complexity of machine learning algorithms, enabling us to gain meaningful insights and accurate predictions while efficiently managing computational resources.

Train / Validation / Test Split:

Once the above data preprocessing and feature reduction steps are completed, our dataset is primed for the exciting world of machine learning algorithms. However, before selecting a specific algorithm, it is essential to divide the dataset into two or sometimes three parts. This separation serves a crucial purpose in the machine learning workflow.

The dataset is typically split into a training set and a validation/testing set. The training set is used to train the machine learning algorithm, allowing it to learn from the available data distribution. The validation/testing set, on the other hand, is employed to assess the algorithm's performance and evaluate its predictive capabilities before deploying it to handle real-world data. By splitting the dataset and performing training, validation, and testing steps separately, we can ensure that the machine learning algorithm is robust and capable of dealing with new and unseen data effectively. This process helps us select the most suitable algorithm that generalizes well and delivers accurate results in real-world scenarios.

Training data: This portion of the dataset serves as the foundation for training machine learning algorithms to build a predictive model. The model endeavors to learn from the training data's various patterns and characteristics, capturing the underlying relationships between input features and target outputs.

Validation data: The validation data segment is utilized to assess and validate different model fits. This process involves fine-tuning model hyper parameters to optimize performance. Unlike the training data, the model does not learn from the validation set directly; instead, it is used to guide the selection and improvement of hyper parameters, leading to a more refined model.

Test data: The test data component is reserved solely for evaluating the model's hypothesis and measuring its performance in a real-world scenario. It remains untouched and unseen until the model and hyper parameters have been finalized using the training and validation data. Afterward, the model is applied to the test data to obtain an accurate measure of its efficacy when deployed on unseen data, providing insights into the model's generalization and real-world performance.

ALGORITHMS:

SVM algorithm:

Support Vector Machine (SVM) is a highly popular Supervised Learning algorithm utilized for both Classification and Regression tasks. However, it is primarily known for its effectiveness in solving Classification problems in the realm of Machine Learning.

The main objective of the SVM algorithm is to construct an optimal line or decision boundary, known as a hyperplane, within the n-dimensional space to effectively segregate different classes. By finding the best hyperplane, the SVM algorithm ensures that new data points can be accurately classified into their respective categories in the future.

In essence, SVM aims to create a robust hyperplane that maximizes the margin between data points of different classes, allowing for a clear and efficient separation. This capability to create an optimal decision boundary makes SVM a powerful and widely used algorithm for various Classification tasks in the field of Machine Learning.

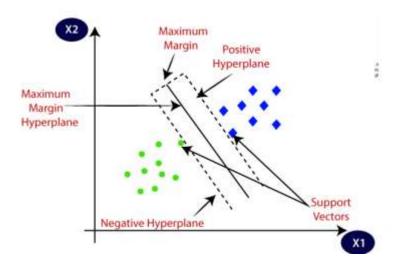


Fig 1.2: Classification using SVM

Important Concepts in SVM:

Support Vectors: Support vectors are data points that lie closest to the hyperplane, which is the decision boundary that separates classes. These support vectors play a crucial role in defining the separating line for different classes.

Hyperplane: The hyperplane is a decision plane or space that divides a set of objects belonging to different classes. It forms the boundary between different classes in the n-dimensional space.

Margin: The margin is the gap between two lines defined by the closest data points of different classes. It represents the perpendicular distance from the hyperplane to the support vectors. A larger margin is considered better, while a smaller margin may lead to a less effective separation.

Advantages of SVM:

Clear Margin of Separation: SVM performs well when there is a distinct margin of separation

between classes, making it suitable for binary classification tasks.

Effective in High-Dimensional Spaces: SVM excels in scenarios with a high number of dimensions, where it can still create efficient decision boundaries.

Suitable for Limited Data Samples: SVM remains effective even when the number of dimensions exceeds the number of samples, making it suitable for various real-world problems.

Memory Efficiency: SVM is relatively memory-efficient, making it scalable to handle large datasets.

Ensemble Model:

An ensemble vote classifier is a machine learning model that combines predictions from multiple individual models to make a final prediction. This approach is based on the concept that aggregating predictions from diverse models can lead to improved overall performance compared to relying on a single model.

In an ensemble vote classifier, each individual model generates a prediction, and the final prediction is determined through a voting scheme. The most common voting schemes are:

Majority Voting: The final prediction is the class that receives the most votes from the individual models.

Weighted Voting: Each individual model is assigned a weight based on its performance on a validation set, and the final prediction is computed as the weighted sum of the individual predictions. Ensemble models, like the vote classifier, are popular for their ability to enhance prediction accuracy, increase robustness, and reduce the risk of overfitting compared to using a single model.

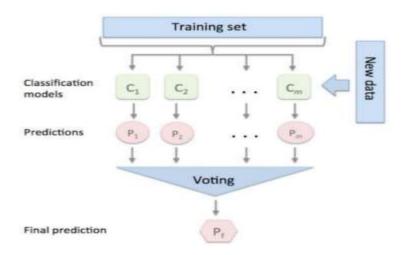


Fig 1.3: Working of Ensemble Model

EXTREME LEARNING MACHINE (ELM) ALGORITHM:

The Extreme Learning Machine (ELM) is a neural network algorithm distinguished by its rapid training speed and impressive generalization capabilities. It functions as a single-layer feedforward neural network. ELM stands out as a single hidden layer neural network, surpassing traditional algorithms in terms of training performance.

One of ELM's key strengths lies in its suitability for handling large-scale datasets. It has been effectively applied to various machine learning tasks, including classification, regression, and feature

extraction.

What sets ELM apart is its automatic implementation, requiring no iterative tuning and eliminating the need for user intervention. The ELM algorithm operates seamlessly without user involvement, making it a powerful and efficient tool for machine learning tasks.

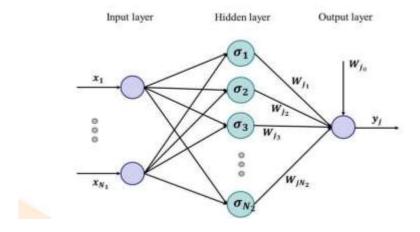


Fig 1.4: Extreme Learning Machine

Existing System:

Invasive Clinical Tests: The traditional clinical evaluation of Inherited Retinal Diseases (IRDs) involves invasive techniques that may not be suitable for infants. These methods may require uncomfortable procedures and can pose risks to the patients.

Sedation of Children: Some clinical tests may necessitate sedation of children, which can have negative effects on their mental health. This approach raises concerns about the well-being and comfort of young patients during the evaluation process.

Computational Time: The existing machine learning approach that utilizes the SVM algorithm for detecting IRDs requires significant computational time for both training and testing phases. This lengthy process may hinder real-time or time-sensitive applications.

Limitations with Larger Datasets: The current approach may encounter challenges when dealing with larger datasets. As the dataset size increases, the SVM algorithm may struggle to efficiently process and analyze the data.

Overfitting Issues: The model developed using the existing machine learning approach may face overfitting problems, especially when the number of attributes exceeds the number of records in the data. Overfitting can lead to reduced predictive accuracy and performance on unseen data.

To address these disadvantages, it is essential to explore alternative approaches that overcome the limitations of the existing system. New methodologies that are less invasive, computationally efficient, and capable of handling larger datasets are required to enhance the clinical evaluation of IRDs and improve patient outcomes.

Proposed System

The proposed system is a machine learning model based on neural networks, specifically using the ELM (Extreme Learning Machine) algorithm, for the detection of inherited retinal diseases (IRDs). This machine learning model allows the system to learn from the training dataset and make informed decisions based on the acquired knowledge.

Advantages of the Proposed System:

IRD Prediction: The system effectively predicts the presence of inherited retinal diseases, aiding in early detection and timely intervention.

Improved Performance: The predictive model's performance is enhanced, leading to more accurate and reliable results in diagnosing IRDs.

Efficient Training and Testing: The training and testing process is streamlined and consumes less time, thanks to the ELM algorithm, which is known for its fast-training capabilities.

Handling Large Datasets: The proposed system demonstrates competence in managing large datasets efficiently, making it suitable for real-world applications with extensive data volumes.

The model is trained on the preprocessed dataset using appropriate data splitting techniques. Additionally, a tkinter-based GUI is being developed, facilitating user interaction with the machine learning model to predict inherited retinal diseases. This graphical interface enhances the system's usability and accessibility, making it a user-friendly tool for medical practitioners and healthcare professionals.



Fig: 1.5 Prediction of diseases

The results "1" represents that the patient is suffering from diseases and "0" represents that the patient has no retinal diseases.



Table 1.1: Comparision Table

	SVM(RIGHT)	SVM(LEFT)	ENSEMBLING	ELM
Accuracy	91.5	94.1	92.8	98.7
Sensitivity	0.91	0.94	0.92	1.0
Specificity	0.73	0.84	0.78	0.85

IV. CONCLUSION

This paper focuses on detecting the presence of inherited retinal diseases (IRDs) using the ELM algorithm. The entire process was carried out on the pupillometric dataset, utilizing accuracy, specificity, and sensitivity as critical evaluation parameters to draw conclusions. A pre-existing system for IRD detection employed an SVM ensemble model, achieving an accuracy of 92.8%. In contrast, our newly developed machine learning model using the ELM algorithm achieved an impressive accuracy of 98.7%. Based on these results, it is evident that the ELM algorithm provides an easy and efficient solution for detecting inherited retinal diseases with pupillometric data. The significantly higher accuracy achieved by the ELM-based model compared to the existing SVM ensemble model showcases the potential and effectiveness of the ELM algorithm for accurate and reliable IRD detection. These findings are valuable for the healthcare sector, as they pave the way for improved early detection and personalized treatment plans for patients with inherited retinal diseases.

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