IJIRCCE

e-ISSN: 2320-9801 | p-ISSN: 2320-9798

INTERNATIONAL JOURNAL OF INNOVATIVE RESEARCH

IN COMPUTER & COMMUNICATION ENGINEERING

Volume 11, Issue 3, March 2023

INTERNATIONAL STANDARD SERIAL NUMBER INDIA

Impact Factor: 8.379

International Journal of Innovative Research in Computer and Communication Engineering

| e-ISSN: 2320-9801, p-ISSN: 2320-9798| www.ijircce.com | |Impact Factor: 8.379 |

|| Volume 11, Issue 3, March 2023 ||

| DOI: 10.15680/IJIRCCE.2023.1103105 |

Automatic Detection of Genetic Disease in Pediatric Age Using Pupillometry

B. Kalyan Chakravarthy[1], Veesam Akshay [2] , Yasam Venkata Srimannarayana[3], Polupalli Suresh [4] , Srungaram Sairam [5]

Associate Professor, Department of IT, Vasireddy Venkatadri Institute of Technology, Nambur, Guntur Dt., Andhra Pradesh, India[1]

UG Students, Department of IT, Vasireddy Venkatadri Institute of Technology, Nambur, Guntur Dt., Andhra Pradesh, India[2,3,4,5]

ABSTRACT : A clinically and genetically diverse category of disorders known as inherited retinal diseases (IRDs) are defined by photoreceptor degradation or malfunction. Children with inherited retinal disorders have severe vision impairments. These are categorised as inner and outer retina illnesses, and they frequently result in childhood blindness. Inborn retinal diseases severely damage the eyesight of children. They frequently cause juvenile blindness and can harm the retina. This type of illness has many clinical and genetic origins, making diagnosis challenging It is typically based on a complex web of invasive clinical tests that aren't always appropriate for babies or young children. Hence, a unique approach utilising chromatic pupillometry is needed, a technique that is increasingly used to gauge how well the outer and inner retina work. This study introduces a Clinical Decision Support System (CDSS) based on deep learning and chromatic pupillometry to facilitate the diagnosis of IRD in paediatric participants.

KEYWORDS: Pediatric age, pupillometry, autonomic nervous system, machine learning, Genetic disease support vector machine.

I. INTRODUCTION

Inherited retinal disorders (IRDs) typically affect children with significant vision deficits.IRDs can be split up into conditions affecting the inner retina, primarily retinal ganglion cell degeneration, and conditions affecting the outer retina, namely photoreceptor degenerations (such as Leber Congenital Amaurosis, Retinitis Pigmentosa, Stargardt diseaseetc). The group of genetic diseases known as retinitis pigmentosa (RP) affect the retina, the light-sensitive layer at the back of the eye. The progressive degeneration of the retina brought on by RP results in loss of vision and eventual blindness.RP is a condition that only affects one in 4,000 people worldwide. Depending on the precise genetic mutation at play, it can be inherited in an autosomal dominant, autosomal recessive, or X-linked pattern.Night blindness is frequently the first sign of RP, which is then gradually there is a loss of peripheral vision and central vision. Some people may continue to have good vision well into their 50s or 60s, while others may go blind legally in their 20s or 30s, depending on the rate of progression.Genetic factors can also be associated with severeness in adults, including glaucoma and macular degeneration The presence of more than 200 causal genes in diseases such as retinitis pigmentosa and other genetic disorders leads to high genetic heterogeneity, which poses significant challenges in accurately diagnosing these conditions. Furthermore, a single gene may lead to multiple clinical presentations, making diagnosis even more difficult. As a result, identifying the specific genetic mutation responsible for a patient's condition can be a time-consuming and challenging process. However, advancements in genetic testing and ongoing research into the underlying causes of these diseases offer hope for improved diagnostic accuracy and more targeted treatments in the future.

The proposed system employs machine learning which uses models like LSTM, BILSTM and Extreme machine learning to predict thegenetic disease inthe individuals during their paediatric age without the need for genetic test or electrophysiological test which saves lot of effort and the expense

| e-ISSN: 2320-9801, p-ISSN: 2320-9798| www.ijircce.com | |Impact Factor: 8.379 |

|| Volume 11, Issue 3, March 2023 ||

| DOI: 10.15680/IJIRCCE.2023.1103105 |

II. LITERATURE SURVEY

[1] Detecting age-related macular degeneration in fundus images
https://dl.acm.org/doi/abs/10.1016/j.compeleceng.2017.11.008
Age-Related Macular Degeneration (AMD) is a severe, degenerative condition that primarily affects adults over 60. Drusen, which are extracellular material deposits in the macular region, are associated with this condition. Finding drusen in fundus pictures is one efficient and non-invasive pre-diagnosis technique for AMD. In this study, we offer a novel approach that integrates mathematical morphology, digital image processing, and a strong machine learning model called a support vector machine (SVM).

[2] Development of machine learning models for diagnosis of glaucoma
https://journals.plos.org/plosone/article?id=10.1371/journal.pone.017772
The goal of the study was to create machine learning models with robust glaucoma prediction capacity and interpretability based on retinal nerve fibre layer (RNFL) thickness and visual field measurements (VF). From measuring the thickness of the retinal nerve fibre layer (RNFL) and the visual field, we gathered a variety of candidate properties (VF). From original features, we also created synthesized features. Afterwards, we chose the top traits that were appropriate for classification.

[3] Creating and deploying a model ensemble is one data analytics approach to creating a clinical decision support system for diabetic retinopathy.
https://www.sciencedirect.com/science/article/abs/pii/S0167923617300908
Diabetes is a typical chronic illness that can cause a number of problems. The most frequent reason for vision loss in diabetic people is diabetic retinopathy (DR), one of these consequences that can be rather devastating. We demonstrate how combining several data preparation and modelling stages aided us in enhancing the functionality of our CDSS.

[4] Fully automated, reliable system to screen optical coherence tomography images for age-related macular degeneration, central serous chorioretinopathy, and retinal edoema
https://www.hindawi.com/journals/bmri/2017/7148245/
Blindness is caused by extensive macula damage, or maculopathy. Age-related macular degeneration (AMD) or retinal edoema (RE) are the main causes (ARMD). The most recent eye testing method that can identify these disorders in their early stages is optical coherence tomography (OCT) imaging.

Problem Identification
Paediatric genetic illnesses can significantly affect children's and families' quality of life. Current diagnostic techniques can be time-consuming, expensive, and intrusive, yet early diagnosis and intervention can be essential in improving results. A non-invasive technique for monitoring pupillary responses, called pupillometry, has showed promise in the early detection of a number of neurological and genetic problems. Its ability to identify hereditary illnesses in paediatric children has not yet been adequately investigated.The goal is to create an automated system using pupillometry for the early detection of genetic diseases in paediatric patients. It should be possible for the system to examine pupillary reactions to visual stimuli and spot aberrant patterns linked to hereditary illnesses. Based on the distinctive pupillary responses of various hereditary illnesses, the system ought to be able to distinguish between them.

Methodology
Thesystem is designed using LSTM, BILSTM and extreme machine learning models.

LSTM
The ability of LSTM to selectively retain or forget information over extended periods of time is its key advantage over conventional RNNs. This is accomplished via a collection of gates and memory cells that manage the information flow inside the network. The gates operatethe flow of data into and out of the memory cells, whilst the memory cells are employed to store data from earlier time steps.

| e-ISSN: 2320-9801, p-ISSN: 2320-9798| www.ijircce.com | |Impact Factor: 8.379 |

|| Volume 11, Issue 3, March 2023 ||

| DOI: 10.15680/IJIRCCE.2023.1103105 |

BILSTM

An expansion of the LSTM architecture used in machine learning, Bidirectional Long Short-Term Memory (BILSTM), processes input sequences both forward and backward. The hidden state at time t in a conventional LSTM network is determined by the current input and the prior hidden state. The hidden state at time t in a BILSTM network is determined from the previous hidden state, the present input, the future hidden state, and the future input. This enhances the network's capacity to represent complicated relationships within the input sequence by enabling it to take into account information from both the past and the future.

Extreme machine learning

ELM is a specific kind of feedforward neural network that uses a single step to solve for the output weights and randomly chooses the weights between the input and hidden layers to train quickly and effectively. ELM is quicker and more effective than standard neural networks because it doesn't require iterative optimisation.

ELM's quick learning pace is one of its key benefits. In comparison to other neural networks, training time is drastically decreased because the weights between the input and hidden layers are randomly assigned. ELM is excellent for big data applications because it can easily manage massive datasets.

Implementation

The system's implementation is divided into four main steps: importing and preprocessing pupillary diameter signals; extracting and reducing pupillary features; optimizing hyperparameters; and, finally, training an extreme machine learning algorithm.

Signal pre-processing:

Signal preprocessing in machine learning entails modifying, transforming, and cleaning input data to make it ready for analysis. The first stage of the CDSS involves analyzing raw files from the binocular pupillometer after each measurement session to export relevant data such as the patient's ID, bilateral pupillary diameter signals, and diagnosis performed by a clinical specialist. Significant clinical features related to pupillary reactivity are extracted from diameter signals and used to build the input dataset for the supervised classifier. During the machine learning system's training phase, the subject's diagnosis is used to label both the subject and their data.However, before extracting the feature set, the raw pupillometric signals must undergo proper processing to reduce noise and address potential eye-blink artifacts that can cause abrupt spikes and reduce the reliability of the pupillary diameter traces.

Feature Extraction

The process of feature extraction in machine learning entails locating and picking out the most crucial features or variables from the raw data. Based on the literature, we identified the most predictive features and extracted eight features from each pupillometric signals after signal pre-processing. These features include maximum and minimum pupil diameter, absolutedifference between them, percentage maximum constriction, latency, mean constriction velocity, maximum constriction velocity, and mean dilation velocity. These features are commonly used in pupillometry research on various pathologies and in biometric authentication, and are also provided by the equipment output files. To ensure accuracy, MAX and LATENCY were calculated in the first second, while the others used a 5-second window. The LATENCY parameter was defined as the delay between the light stimulus and the onset of pupillary constriction, estimated by computing the first derivative of the pupillometric signal and identifying the time instant corresponding to $d(t) = 0$ by checking backwards from its absolute minimum, without identifying inflection points due to the presence of significant noisy components.

Feature Reduction

Feature reduction in machine learning eliminates multicollinearity, which improves the current machine learning model.

A total of 292samples were extracted from the 36 pupil reactivity signals, making them available for classification Due to the relatively large number of features, feature reduction was a crucial first step that was used to prevent the training dataset from becoming overfit. Keep the dimension of the input feature space for machine learning applications below one

| e-ISSN: 2320-9801, p-ISSN: 2320-9798| www.ijircce.com | |Impact Factor: 8.379 |

|| Volume 11, Issue 3, March 2023 ||

| DOI: 10.15680/IJIRCCE.2023.1103105 |

Feature	Description	Expression
MAX	maximum diameter at baseline	$MAX(r(t))$
MIN	minimum diameter corresponding to the peak constriction	$MIN(r(t))$
DELTA	difference between Max and Min	$MAX\text{-}MIN$
CH	percentage maximum constriction	$\frac{DELTA}{MAX}$
LATENCY	delay between stimulus and onset of the pupillary constriction	Computed using custom script
MCV	mean constriction velocity	$\frac{DELTA}{t_{min}-LATENCY}$
MDV	mean dilation velocity	$\frac{r(t)_{80\%}}{t_{80\%}-t_{min}}$
CV_{max}	maximum constriction velocity	$MIN\left(\frac{dr(t)}{dt}\right)$

Feature calculation
fifth of the total number of observations, or the best subjects. The set of features used in this investigation is based on the findings of a prior study that found a subset of pupillary features to have greater discriminating power for the clinical diagnosis of RP

Model training
Extreme Learning Machine (ELM) is a machine learning algorithm that is widely used for classification and regression tasks. The training and testing data for the right and left features, as well as the corresponding labels, are first concatenated which are obtained from the data pre-processing.
The E-ELM classifier is then trained using the concatenated data using a specific variant of the ELM algorithm known as the Generalized ELM classifier (GenELMClassifier).This process creates a hidden layer for the ELM model using the MLPRandomLayer class, which stands for Multi-Layer Perceptron Random Layer. This layer will be used to transform the input features into a higher-dimensional space, where the ELM model can more easily learn a decision boundary between different classes.The activation_func parameter is used to specify the activation function of the neurons in the hidden layer. In this case, the hyperbolic tangent function (tanh) is used.

Classification
Classification involves several steps it creates the ELM classifier using the GenELMClassifier class, which implements the generalized ELM algorithm.Once the ELM model is trained, it can be used to make predictions on new data using the predict method of the GenELMClassifier object. After fitting the classifier to the concatenated training data, it proceeds to predict the labels for the concatenated testing data using the prediction function. The accuracy of the E-ELM classifier is then evaluated by comparing the predicted labels to the true labels in the testing data using the accuracy_score function.

IV. RESULTS

User Inputs:

International Journal of Innovative Research in Computer and Communication Engineering

| e-ISSN: 2320-9801, p-ISSN: 2320-9798| www.ijircce.com | |Impact Factor: 8.379 |

|| Volume 11, Issue 3, March 2023 ||

| DOI: 10.15680/IJIRCCE.2023.1103105 |

Output

prediction result if there is chance of disease

Fig.8. Prediction Result if there is no disease

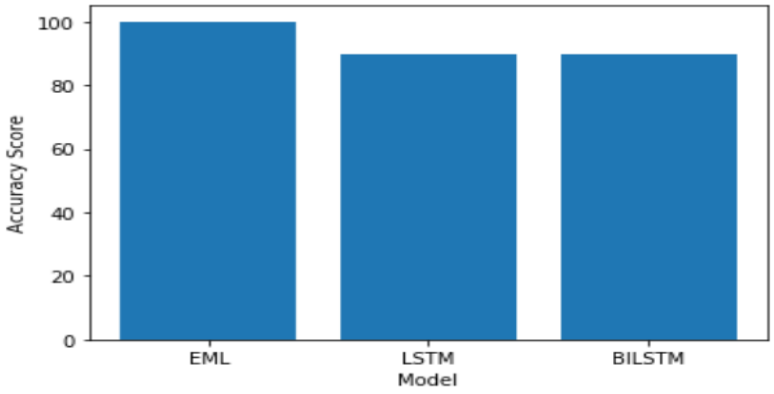

Fig.9. Accuracy Bar Graph

V. CONCLUSION

In conclusion, the Performances were assessed separately for the right and left eyes using a leave-one-out cross-validation, which was also used to determine the ideal configuration of the SVM's internal parameters. In order to increase the overall sensitivity of the CDSS, the class assigned to each eye was ultimately combined with an OR-like methodology;, the Extension Extreme Learning, SVM on the right eye (accuracy is 89%), SVM on the left eye (accuracy is 12%), Ensemble on both eyes (accuracy is 11), LSTM (accuracy is 89%), and BILSTM (accuracy is 89%).Further Extension using extension extreme machine learning algorithm. It's a training algorithm for a single hidden layer feed-forward neural network that we achieved accuracy to 99 percent and more improvement over sensitivity and specificity.

Future Scope

A promising area of research with potential future healthcare applications is the automatic detection of RP in children. This will likely become more accurate and effective as machine learning algorithms and artificial intelligence develop.

| e-ISSN: 2320-9801, p-ISSN: 2320-9798| www.ijircce.com | |Impact Factor: 8.379 |

|| Volume 11, Issue 3, March 2023 ||

| DOI: 10.15680/IJIRCCE.2023.1103105 |

The outcomes and quality of life for those who are affected and their families may improve as a result of earlier and more accurate diagnosis of genetic diseases in children.
The development of individualised treatment plans for each patient can be facilitated by the use of this technology to track the development of genetic diseases and the efficacy of treatment. In the end, this may result in more precise and effective treatment and better health outcomes.

REFERENCES

1. D. T. Hogarty, D. A. Mackey, and A. W. Hewitt, ‘‘Current state and future prospects of artificial intelligence in ophthalmology: A review,’’ Clin. Exp. Ophthalmol., vol. 47, no. 1, pp. 128–139, Jan. 2019.
2. R. Kapoor, S. P. Walters, and L. A. Al-Aswad, ‘‘The current state of artificial intelligence in ophthalmology,’’ Surv. Ophthalmol., vol. 64, no. 2, pp. 233–240, Mar. 2019.
3. N. Brancati, M. Frucci, D. Gragnaniello, D. Riccio, V. Di Iorio, L. Di Perna, and F. Simonelli, ‘‘Learning-based approach to segment pigment signs in fundus images for Retinitis Pigmentosa analysis,’’ Neurocomputing, vol. 308, pp. 159–171, Sep. 2018.
4. S. S. Gao, R. C. Patel, N. Jain, M. Zhang, R. G. Weleber, D. Huang, M. E. Pennesi, and Y. Jia, ‘‘Choriocapillaris evaluation in choroideremia using optical coherence tomography angiography,’’ Biomed. Opt. Exp., vol. 8, no. 1, p. 48, Jan. 2017.
5. García-Floriano, Á. Ferreira-Santiago, O. Camacho-Nieto, and C. Yáñez-Márquez, ‘‘A machine learning approach to medical image classification: Detecting age-related macular degeneration in fundus images,’’ Comput. Electr. Eng., vol. 75, pp. 218–229, May 2019.

INNO SPACE
SJIF Scientific Journal Impact Factor

Impact Factor: 8.379

doi cross ref

ISSN INTERNATIONAL STANDARD SERIAL NUMBER INDIA

निस्केयर NISCAIR

INTERNATIONAL JOURNAL OF INNOVATIVE RESEARCH

IN COMPUTER & COMMUNICATION ENGINEERING

9940 572 462 6381 907 438 ijircce@gmail.com

www.ijircce.com

Scan to save the contact details