VARDHAMAN COLLEGE OF ENGINEERING
(AUTONOMOUS)
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Team No. 2020040103

Project Work Phase-II (A6445)
Domain: Communication

Multi-Algorithmic Retinal Genetic Disease Detection using Pupillometry

By

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Outline:

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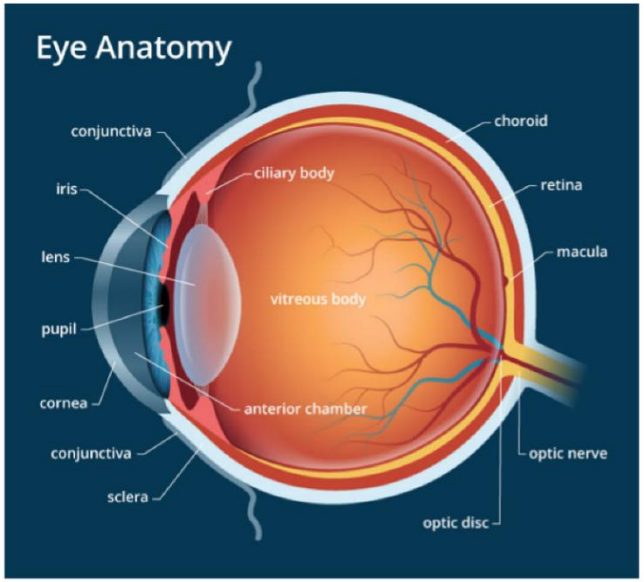
Introduction:

- Retinitis Pigmentosa (RP) is a group of inherited retinal disorders characterized by progressive vision loss.
- RP can manifest either during childhood (early onset RP) or adulthood, impacting individuals across various age groups.
- The primary symptom of RP is night blindness, where individuals experience difficulty seeing in low-light conditions.
- As the condition progresses, RP leads to the development of blind spots in the peripheral (side) vision.
- Over time, these blind spots expand, resulting in reduced peripheral vision and, in severe cases, tunnel vision or total blindness.

Fig: Pupillometric Device

Motivation:

- The motivation lies in the fact that genetic diseases are often difficult to detect in children, especially in their early stages.
- This can lead to delayed diagnoses and treatments, which can have serious consequences for the child's health and well-being.
- By implementing various algorithms, we can reduce the time and cost associated with traditional diagnostic methods, such as blood tests and genetic screenings.



Literature Survey:

Title	Automatic Detection of Genetic Diseases in Pediatric Age Using Pupillometry
Journal Name and Year	IEEE International Conference for Convergence in Engineering and Year: 2020
Authors	Ernesto Iadanza, Francesco Goretti, Michele Sorelli, Paolo Mellilo
Contribution	This paper presents a novel Clinical Decision Support System (CDSS), based on Machine Learning using Chromatic Pupillometry to support diagnosis of inherited retinal diseases in pediatric subjects.
Parametric Analysis	Accuracy= 0.846 , Sensitivity=0.937, Specificity= 0.786

Literature Survey:

Title	DNA Diagnosis in Case Series of Hereditary Retinal Dystophy
Journal Name and Year	IEEE Journal on Cognitive Sciences, Genomics and Bioinformatics and Year: 2020
Authors	Tatyana Vasilyeva, Vitaly Kadyshev, Andrey Marakhonov, Rena Zinchenko
Contribution	Diagnoses were made based on ophthalmologic examination, including best-corrected visual acuity, slit-lamp biomicroscopy, ophthalmoscopy, and fundus examination, electroretinograms, and Goldmann visual field measurements, as well as based on the pedigree analysis
Parametric Analysis	NGS serves an indispensable method for DNA diagnostics of rare forms of inherited retinal dystrophy which have a remarkably similar clinical picture at an advanced stage.

Problem Statement:

- The problem with traditional methods of detecting genetic diseases in children is that they can be invasive, time-consuming and expensive.
- For instance, a blood test may require multiple samples from the child, and it may take weeks or months to get results.
- By measuring changes in the size of the pupil in response to light, pupillometry can provide valuable insights into a child's health without the need for invasive procedures.

Objective:

- Develop a comprehensive multi-algorithmic approach for the detection of retinal genetic diseases, with a focus on Retinitis Pigmentosa (RP).
- Provide a versatile tool for healthcare professionals to accurately diagnose Retinitis Pigmentosa, facilitating timely interventions and personalized treatment plans.

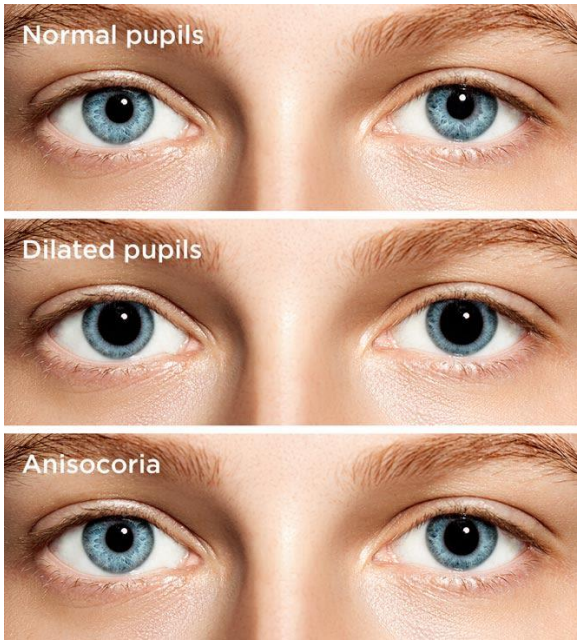
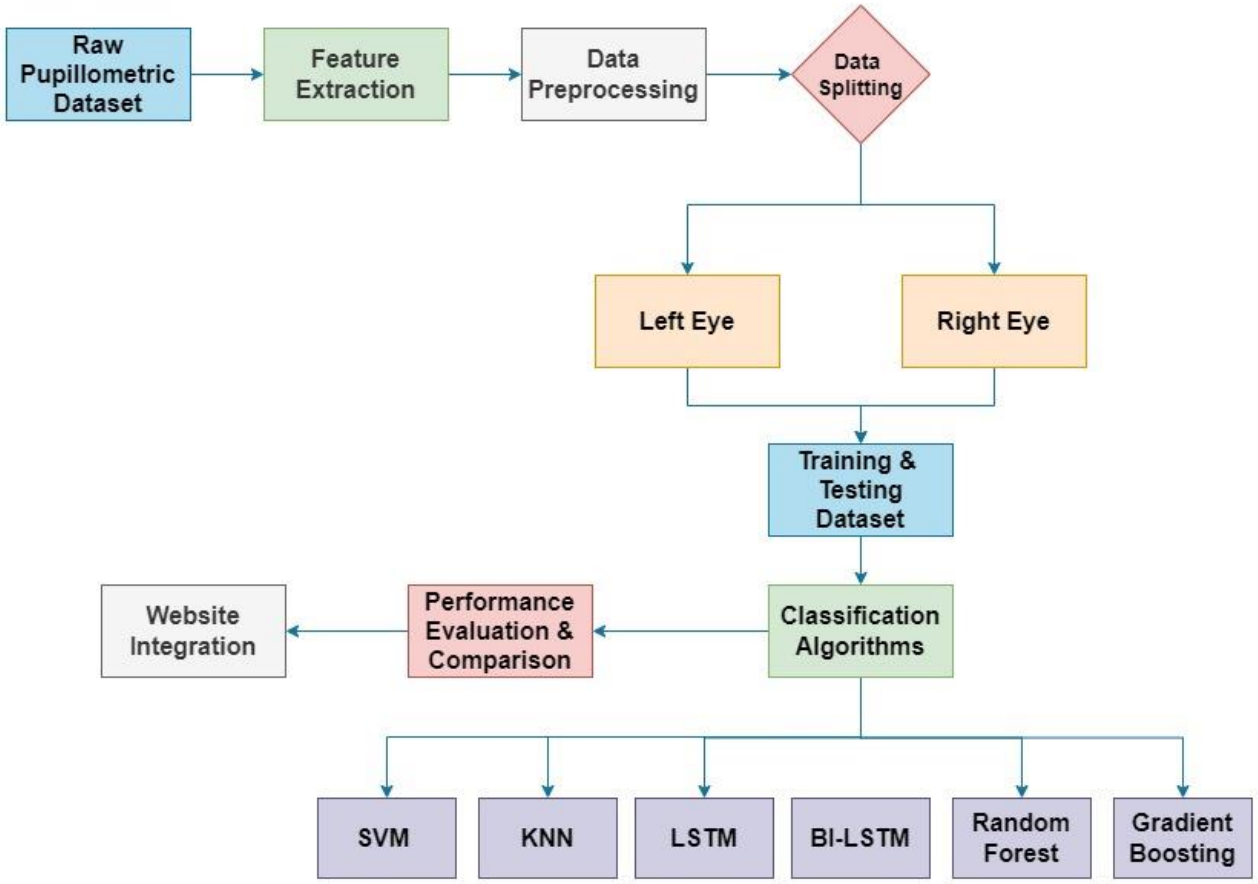


Fig: Different pupil sizes

Block Diagram:



Methodology:

➢ Random Forest:

It begins by splitting the dataset into training and testing sets. Then, the algorithm is instantiated with specified hyperparameters like the number of decision trees (estimators) and maximum depth. Next, the Random Forest classifier is trained on the training data using the 'fit' method. After training, predictions are made on the testing data using the predict method

➢ Gradient Boosting:

In the Gradient Boosting algorithm implementation, it starts by preprocessing the dataset, splitting it into training and testing sets. Then, the Gradient Boosting Classifier is instantiated with specified hyperparameters like the number of estimators, learning rate, and maximum depth. Next, the classifier is trained on the training data using the 'fit' method. Following training, predictions are made on the testing data using the predict method.

KNN:

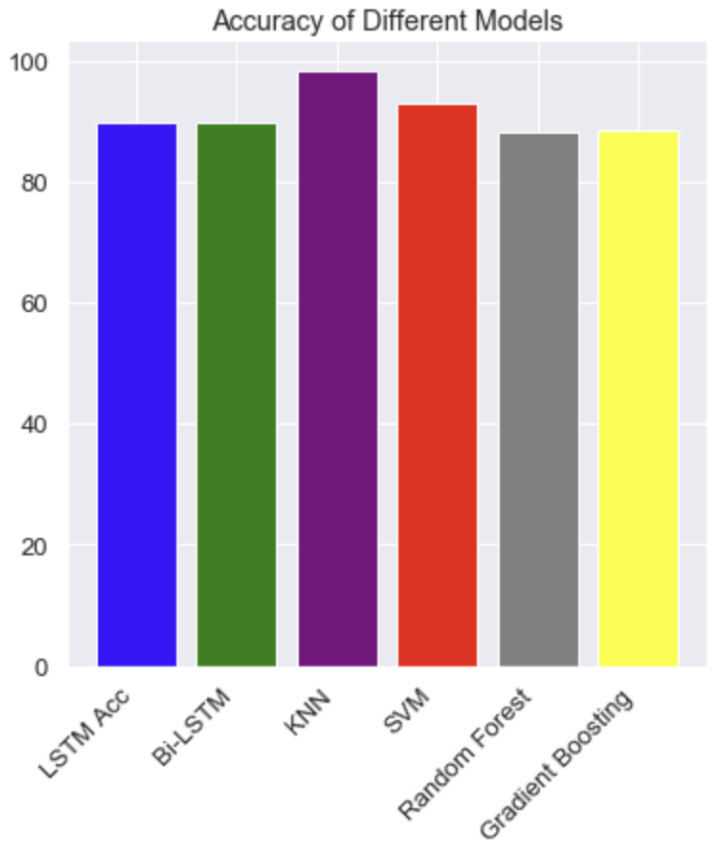
It begins by combining training and testing datasets. During training, the algorithm stores the feature-label mappings. To make predictions, it computes the distance between each test instance and all training instances, then selects the K nearest neighbors. The class label with the highest frequency among these neighbors is assigned to the test instance. Finally, the algorithm evaluates its accuracy by comparing predicted and actual labels.

LSTM:

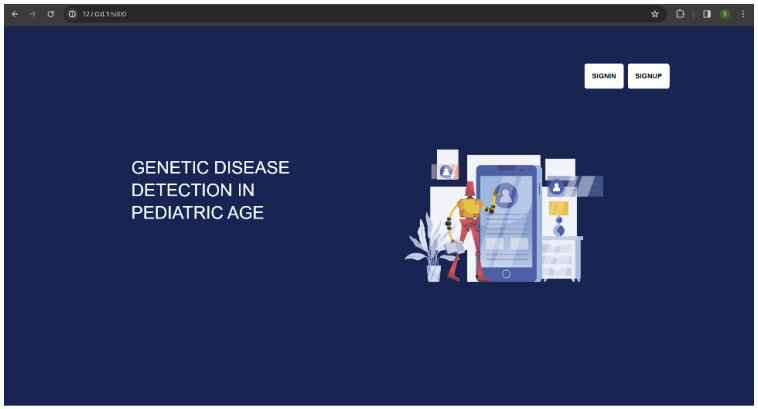
The algorithm preprocesses the target variable Y using one-hot encoding to convert categorical data into a binary matrix format. It reshapes the input features X to be compatible with LSTM input requirements. Sequential model architecture is defined with LSTM layers for sequence processing, followed by dropout to prevent overfitting and dense layers for classification. The model is compiled with binary cross-entropy loss and Adam optimizer. Training commences, and after completion, model accuracy is evaluated. Finally, predictions are made using the trained model.

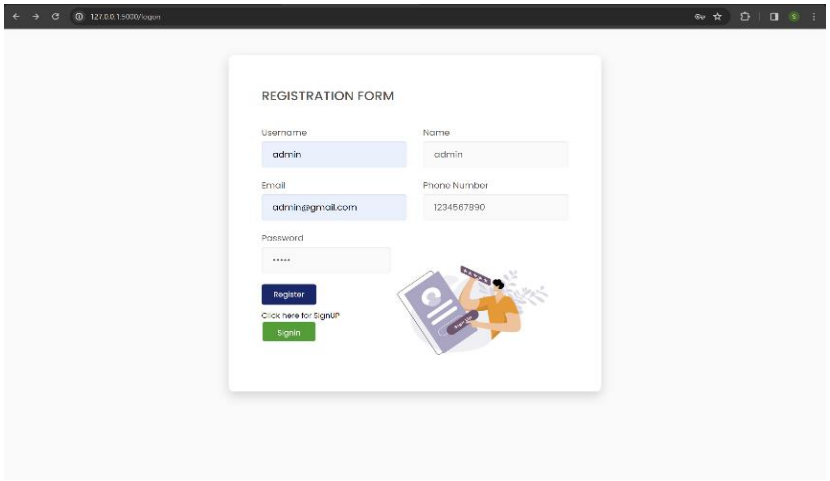
Results:

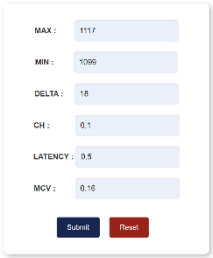
SVM Accuracy: 92.85
KNN Accuracy: 98.31
LSTM Accuracy: 89.6
Bi-LSTM Accuracy: 89.8
Random Forest Accuracy: 88.18
Gradient Boosting Accuracy: 88.43



Output Screens:







Conclusion:

- The multi-algorithmic approach demonstrates promising results for retinal genetic disease detection.
- By leveraging a combination of machine learning and deep learning techniques, we achieved high accuracy rates.
- This comprehensive approach shows potential for robust clinical applications in early disease diagnosis.

References:

[1] E. Iadanza *et al.*, "Automatic Detection of Genetic Diseases in Pediatric Age Using Pupillometry," in *IEEE Access*, vol. 8, pp. 34949-34961, 2020.

[2] S. Rafique, N. Kanwal, I. Karamat, M. N. Asghar and M. Fleury, "Towards Estimation of Emotions From Eye Pupillometry With Low-Cost Devices," in *IEEE Access*, vol. 9, pp. 5354-5370, 2021.

[3] T. Vasilyeva, V. Kadyshev, A. Marakhonov and R. Zinchenko, "DNA Diagnosis in Case Series of Hereditary Retinal Dystrophy," *2020 Cognitive Sciences, Genomics and Bioinformatics (CSGB)*, Novosibirsk, Russia, 2020, pp. 101-105.

Any Questions/Discussions ???

Thank You !!!