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# Automatic Detection of Genetic Diseases in Pediatric Age Using Pupillometry

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***Abstract*— Inherited retinal diseases cause blindness in children. They are classified as wet macular degeneration and dry macular degeneration, which are caused by blindness in children. The diagnosis for these diseases is difficult; it is based on a complex pattern of clinical tests that are inappropriate for infants. So, there is a need for a different approach that utilizes chromatic pupillometry, which is used to access retinal disorders. This paper presents a different approach for detecting retinal diseases in pediatric age that uses a clinical decision support system (CDSS) based on machine learning using chromatic pupillometry to support the diagnosis of inherited retinal diseases in pediatric age. One approach is to use a pupillometer to collect the data. Two different support vector machines—one for the left eye and another for the right eye—classify the features extracted from pupillometer data. The designed clinical decision support system is used for diagnosis. The results are obtained by combining two SVMs in an ensemble model.**

**Keywords— Retinal diseases, Chromatic pupillometry, Clinical Decision Support System, Machine learning, Pupillometer, Support vector machine, RP, OCT, MCV, MDV.**

## I. INTRODUCTION

Automatic Detection of Genetic Diseases in Pediatric Age using Pupillometry describes the concept of detecting eye genetic diseases in pediatric age using pupillometry device data, as this device is very accurate and does not require a huge number of clinical tests to detect disease. All existing techniques require a huge number of clinical tests to diagnose eye pupil disease in children, and it's not good for the children's health. So, using a pupillometry device that captures pupil diameters continuously and records that data in raw format in the file. The use of chromatic pupil responses may be a novel way to diagnose and monitor diseases affecting either the outer or inner retina[1]. Later, we can analyze that data using the machine learning SVM algorithm to detect the presence of disease. Here, using two different SVM classifiers to train right and left eye pupil data, and then performing OR operations between the two classifiers using the ensemble classifier to get a classifier with better accuracy, SVM will predict disease if the pupil's size is huge.

## II. LITERATURE SURVEY

When it comes to systems for "rare diseases," "retinitis pigmentosa," and "pupillometry," the number of studies falls. In order to research individuals with RP, Brancati et al.[10] use machine learning-supervised algorithms for detecting pigment indications on fundus images captured with a digital retinal camera. By identifying intact choriocapillaris, Gao et al.[2] use the ML random forest technique on optical coherence tomography (OCT) images to support the diagnosis of choroideremia. Four further publications use comparable supervised ML algorithms to study common eye conditions like glaucoma[3], diabetic retinopathy[4], and age-related macular degeneration[5].Indeed in previous works the authors successfully used ML for creating CDSSs related to chronic diseases such as congestive heart failure[8] or chronic obstructive pulmonary disease[9].

## III. IMPLEMENTATION

A machine learning SVM algorithm is selected as supervised classification algorithm due to its proven solidity and versatility for classification problems [7] and used to detect the presence of the disease. Pupillometric data is collected to evaluate the retinal and optical nerve function. Pupillometry data is a set of measurement values of pupillary response to light stimuli. The raw pupillometric signals must be processed to attenuate noisy components, to cope up with potential eye-blinks which corrupt the resultant traces of pupil diameter. For this, we use Savitzky-Golay of third-order smoothing filter [6], a digital filter.The 8-elements vector of features is extracted from the pupillometric signal .MAX, MIN, DELTA, CH, LATENCY, MCV, MDV, and CVmax [8] calculate on the filtered signal. A total of 288 features was

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extracted from the 36 pupil reactivity signals, available for each subject to be classified. Due to the relatively high number of features, feature reduction represented a key preliminary operation applied to avoid overfitting the training dataset.

The support vector machine algorithm is used to create hyperplane that can segregate n-dimensional space into classes so that we can easily put the new data point in the correct category.

The system is designed to separately label the left and right eyes and then to classify the related subject by means of an OR logical operator, i.e., the subject is Retinas Pigmentosa detected if at least one of the eyes is assigned with "pathologic" label.

### A. *Abbreviations and Acronyms*

SVM: Support Vector Machine

CDSS: Clinical Decision Support System

RP: Retinitis Pigmentosa

OCT: Optical Coherence Tomography

MCV: Mean Constriction Velocity

MDV: Mean Dilation Velocity

### B. *Figures and Tables*

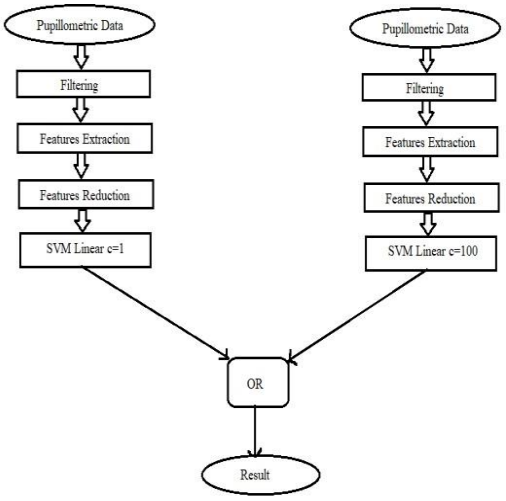


Fig. 1. Architecture of the system

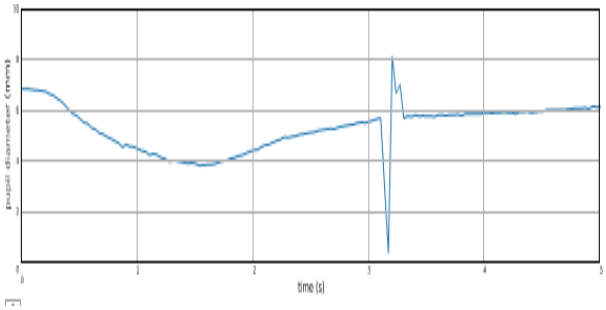


Fig. 2. shows the data analysis, selection of features and optimization of the SVM parameters.

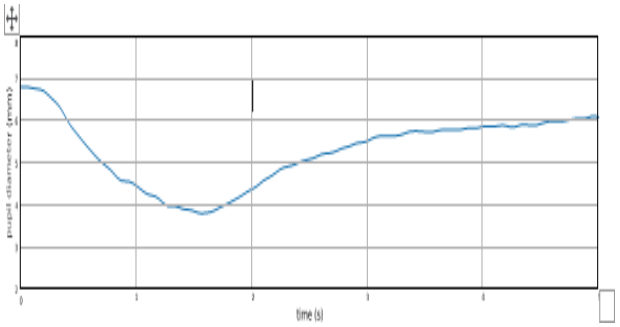


Fig. 3. example of filtered signal


Fig. 4. Upload pupillometry data

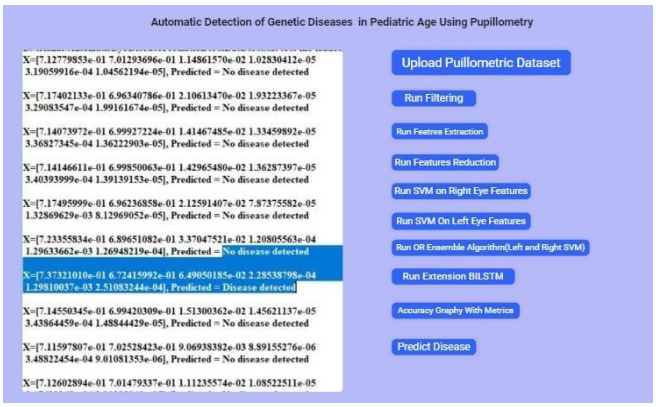


Fig. 5. shows the data analysis , selection of features and optimization of the SVM parameters

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TABLE I. Pupillary reactivitty: extracted features.

| 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 | $MAX - MIN$ |
| 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 | $MIN(\frac{dr(t)}{dt})$ |

## IV. CONCLUSION

This paper describes a new approach for supporting clinical decisions for the diagnosis of retinitis pigmentosa starting from the analysis of pupil response to chromatic light stimuli in pediatric patients. The system was developed to clean artefacts, extract features, and help with the diagnosis of RP using a ML approach based on an ensemble model of two fine-tuned SVMs. Performances were evaluated with leave-one-out cross-validation, also used to identify the best combination of internal parameters of the SVM, separately for both the left and right eyes. The classes assigned to each eye were combined in the end with an OR-like approach to maximize the overall sensitivity of the CDSS; the ensemble system achieved 84.6% accuracy, 93.7% sensitivity, and 78.6% specificity. The small amount of data available for this work calls for further tests with a larger data pool for validating the performance of the system. Future scope includes testing the same approach with different devices.

## ACKNOWLEDGMENT

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