

Autoencoder-based Gaussian mixture model for diagnosing early onset of diabetic retinopathy

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ABSTRACT

The current study presents a simplified yet innovative solution towards effective early diagnosis of diabetic retinopathy (DR) that leads to irreversible blindness. A review of current literature shows a considerable number of machine learning and deep learning approaches have been presented; however, there are significant issues with the early detection of DR. Hence, the proposed study deploys a novel architecture using an autoencoder that extracts a hidden representation of retinal images while binary classification is carried out using a Gaussian mixture model. The prime contribution is the joint integration of deep learning with statistical modelling towards efficient feature extraction and anomaly detection, supporting early determination of DR. The study outcome shows a proposed system to significantly exhibit 96.5% accuracy, 94.2% sensitivity, and 98.3% specificity on two standard benchmarked datasets in comparison to existing models frequently used for the diagnosis of DR.

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1. INTRODUCTION

Diabetic retinopathy (DR) is considered one of the critical medical complications of diabetes that leads to irreversible vision loss [1]. The progression of DR is characterized by various stages, right from mild score to very high score of severity. This progression can be slowed down by vitrectomy, anti-VEGF injection, laser therapy, etc. However, the most challenging part of this medical condition is that it is very problematic to identify in its early stages owing to its asymptomatic form. This is the major reason that the majority of conventional screening for ocular diseases fails to identify DR in its early stages. There are various screening methods for DR in current times in modern clinical setups, viz., fundus photography, examination of dilated fundus, optical coherence tomography (OCT), fluorescein angiography and automated retinal screening system [2]. All these existing screening approaches are non-invasive with high-resolution, detailed images; however, there are challenges associated with almost all of them. For example, fundus photography and direct ophthalmoscopy are not suitable for patient who has other ocular conditions, and they demand pupil be dilated during examination (which may not be suitable for many patients). OCT demands highly specialized devices and yet they have limitations in detecting the early onset of DR until and unless there are no potential changes observed in the macular or retinal area. Fluorescein angiography is invasive and therefore it cannot be expected to be used routinely for patients. Automated retinal screening system suffers from unavoidable outliers and demand consistent updates, while there is a higher cost of equipment for retinal scanning technology. As a part of the evolving solution, it is noted that artificial intelligence (AI) has been a significant contributor towards medical image processing using its machine learning (ML) and

deep learning (DL) algorithms [3]. The adoption of AI models can be used for analyzing retinal images to identify various abnormalities and hence diagnostic process is accelerated while AI minimizes its dependency on human experts. Healthcare providers are assisted with various valuable clinical suggestions based on data analyzed for better and faster decision-making. Hence, AI offers increased efficiency, reduced human error, and remote screening towards a diagnosis of DR. Further, ML is a subset of AI and formulates decisions by learning patterns of screened data. The retinal images can be classified by ML based on different stages of DR while it also facilitates differentiating DR from other ocular diseases. Hence, ML contributes to enhanced prediction of risk with improved diagnosis, scalability, and personalized care. Finally, DL is an advanced subset of the ML algorithm that uses many layers to increase the learning capability for gaining complex insights. Free from any manual extraction of features or preprocessing, retinal images can be directly subjected to DL. DL is capable of autonomously identifying the appropriate features e.g., haemorrhage, exudates, changes in blood vessels, etc. that could reflect the positive case of DR. Various advanced levels of multi-category classification can be carried out by DL offering much granularity in disease progression. Hence, the DL model contributes to higher accuracy, early detection, and automation of workload.

However, there are challenges too associated with AI, ML, and DL which are mainly associated with data quality, interpretability, and overfitting issues. There are still various contradictory theories to claim whether ML or DL could prove fruitful towards the early detection of DR in real-time scenarios.

2. RELATED WORK

Various related works have been studied to understand different variants of methods used for diagnosing DR [4]-[12]. The first frequently used learning model is the convolutional neural network (CNN) which is proven to have higher adoption in addressing classification tasks related to medical images. Various existing research works using CNN have classified retinal images for screening DR [13]-[17]. The second widely-used machine learning model for screening DR is the support vector machine (SVM) to find its higher side of adoption towards the feature-based classification of retinal images [18]-[21]; however, they are found to be less effective when subjected to complex patterns of DR images. The next frequent adoption is noted for the deep belief network (DBN) which is a type of deep learning approach where the Restricted Boltzmann Machine is used for developing its multiple layers so that hierarchical feature learning can be carried out. At present, there are various evolving studies adopting DBN towards training extracted features for the detection of DR [22]-[24]. ResNet-50 is another widely used approach for DR screening which is a type of deep residual network that is capable of addressing vanishing gradient problems. Current studies have been witnessed to adopt ResNet-50 increasingly for its capability of increased accuracy and higher specificity performance [25]-[28].

After reviewing existing studies, the following research problems have been identified: i) early-stage detection: Existing state-of-the-art deep learning methods and other associated approaches encounter potential challenges in differentiating early signs of DR from normal images of the retina. ii) inadequate generalization over diverse data: Existing methods like ResNet-50 and CNN, claimed to be potentially strong were found to overfit on the particular dataset and hence they exhibit much poor generalization when assessed on a different dataset, iii) Complexity of learning approach: The complex architectures like ResNet-50 demand significant computational resources (for both inference and training) acting as an impediment towards deployment in presence of environment with resource-constraints. iv) Sub-optimal interpretability of deep learning model: Although ResNet-50 and CNN are known for higher accuracy, their outcome is less easily interpretable for adopting a black box model in their underlying architecture.

The prime aim of the proposed study is to develop a simplified yet innovative diagnosis framework for the detection and classification of DR at its early onset. The value-added contribution of the study are as follows: i) A novel autoencoder-based architecture has been presented to diagnose DR in early stages with higher accuracy, ii) the study models facilitate identification of abnormalities as DR indicators using Gaussian mixture models (GMM) and reconstruction errors, iii) an exhaustive assessment is carried out to compare proposed model with CNN, SVM, and ResNet-50 on two widely used standard benchmarked dataset, and iv) a potential generalization is demonstrated by the model over diverse dataset which shows its applicability on near real-world screening of DR.

3. METHOD

The prime objective of the proposed research work is towards evolving a novel deep learning model that can facilitate potential learning of hidden representation of retinal images for early detection of DR. The proposed study has coupled a novel autoencoder with simplified mathematical modelling to identify as well as classify the abnormalities associated with the retina during DR. An explicit research method with a

sequential and progressive operational step has been performed to accomplish this study goal. Figure 1 highlights the architecture of the proposed system.

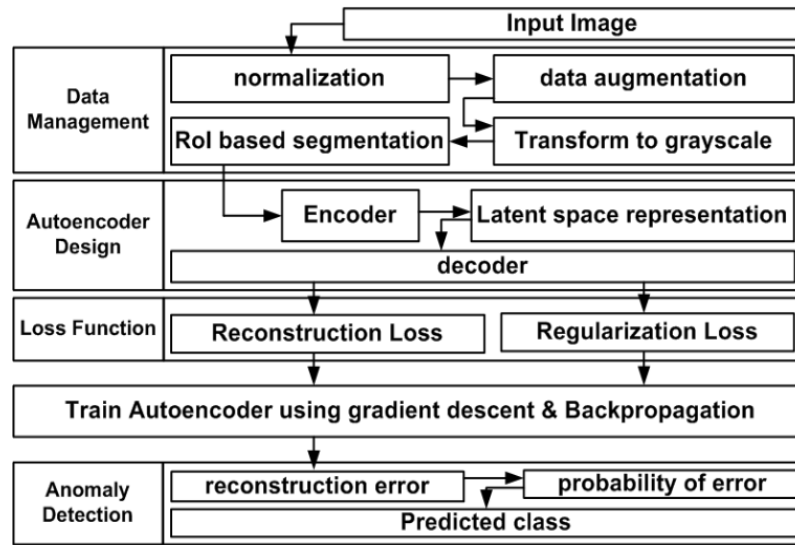


Figure 1. The architecture of the proposed system

According to Figure 1, there are various sets of operations being carried out by the proposed system which start from the management of the dataset to obtaining the outcome of the study. The method uses reconstruction errors to find anomalies suggestive of early-stage diabetic retinopathy (DR) by combining autoencoders with Gaussian mixture models in a novel way. To the best of our knowledge, the integration of autoencoders and GMM into a single framework specifically for early DR detection has not been investigated, even though both components have been employed independently in the literature to date. This hybrid technique increases the sensitivity of early-stage detection by enabling both feature extraction (using autoencoders) and classification (using GMM). Following is the elaborated discussion of the operational process being undertaken in the proposed system:

3.1. Data management

The input image is subjected to standardization where rescaling is performed on all retinal images to fixed $W \times H$ pixels, which is an essential step towards ensuring proper compatibility of the input image to the architecture of the neural network. Consider that, the retinal image is represented as X where $X \in R^{W \times H \times C}$, while the variable C represents the cardinality of colour channels. A simple way to understand this is that $C=1$ will represent a grayscale image while $C=3$ will represent an RGB image. The mathematical representation of normalization is as,

$$X_{norm} = \frac{A^1}{\sigma} \quad (1)$$

In (1), the computation of normalized image X_{norm} is carried out by dividing variable A^1 by standard deviation σ , where A^1 represents a difference of mean μ from main pixel X i.e. $A^1 = X - \mu$. After obtaining a normalized image, data augmentation is performed to maximize the variability and size of the dataset. The mathematic representation of data augmentation is as:

$$X' = R_{\theta}(X) \quad (2)$$

In (2), X' represents augmented data while R_{θ} represents rotation transformation considering θ as random rotation. Apart from this, images could be transformed into grayscale images if the model is anticipated to focus more on texture and share despite colour attributes. The study will perform segmentation using region-of-interest (RoI) to confine its attention to the retinal area and eliminate any unnecessary regions in the background.

3.2. Autoencoder design

The proposed study will use a convolution autoencoder (CAE) which is designed using three core components i.e., encoder, latent space representation, and decoder. The encoder extracts feature maps from the input image using the activation function, pooling layers, and convolution layers. The latent space representation is an outcome from a prior encoder that captures all potential features from the retina e.g., microaneurysms, exudates, and blood vessels. The decoder performs reconstruction of the final image using transposed layers of convolution from the prior module i.e. latent space. The mathematical representation of the encoder operation is as follows:

$$Z=E(X) \quad (3)$$

In (3), the variable Z represents the latent representation of the input image where E represents an encoder function. The expression can be further modified as $E(X)=f_{\theta_1}(X_1)$ where E is the encoding function f_{θ_1} represents the rectified linear unit (ReLU) function while X_1 represents the first convolution operation. The convolution is applied in each subsequent layer followed by activation function and pooling. The input image X is transformed to Z , a latent representation, after passing via different layers. It should be noted that Z , a latent vector, is a highly compressed form of an image with low dimension while the feature extraction capability of the autoencoder is decided by d i.e., the dimensionality of Z latent vector. In the final step, the decoding operation is applied which is mathematically represented as,

$$\hat{X} = D(Z_1) \quad (4)$$

In (4), the variable \hat{X} Represents a reconstructed image while D represents the decoding function using the same activation and Z_1 represents transposed convolution operation. It should be noted that the prime purpose of this CAE is towards learning a representation of both normal and DR images followed by input image reconstruction using low-dimensional features. Any form of anomalies presented in the form of errors during the reconstruction process can offer indicators of early DR. The novelty of this CAE module different from any existing system is i) This scheme uses an attention mechanism to emphasize RoI (e.g. exudates, microaneurysms) that are clinically proven to be critical for the diagnosis of DR and ii) The study also uses dilated convolutions (also known as variational autoencoders (VAE)) to manage features of retinal images with higher complexities as well as fluctuation in stages of DR.

3.3. Loss function

The prime notion of the autoencoder is towards reduction of all possible scores of reconstruction error between the X and \hat{X} , i.e., input and reconstructed image. The study uses mean squared error (MSE) as a loss function in the initial rounds of operation. It is empirically expressed as,

$$L_{recon} = \frac{1}{N} \sum_{i=1}^N ||X_i - \hat{X}_i||^2 \quad (5)$$

In (5), the computation of loss due to reconstruction L_{recon} is carried out considering N as the number of images while the original and reconstructed image is represented as X_i and \hat{X}_i Respectively. Apart from this, the proposed scheme also uses L2 regularization to control the overfitting problem. The mathematical representation of loss due to regularization is shown as,

$$L_{regu} = \lambda \sum_j ||\theta_j||^2 \quad (6)$$

In (6), the loss of regularization L_{regu} is computed considering network weights as θ_j and the constant of regularization as λ . It should be noted that the idea of using L2 regularization is to induce a penalty towards large weights present within the network. Hence, the total loss function L_{total} is computed by adding (5) and (6) i.e. $L_{total}=L_{recon}+L_{regu}$.

3.4. Anomaly detection using reconstruction error

After the computation of the loss function, the proposed system uses backpropagation and gradient descent for training the autoencoder. Assuming the network parameter be denoted by $\theta=\{\theta_1, \theta_2\}$, where θ_1 and θ_2 represent both encoder and decoder respectively. Hence, the update rule can be defined as:

$$\theta \leftarrow \theta - \eta \Delta_{\theta} . L_{total} \quad (7)$$

In (7), the learning rate is signified as η while $\Delta_{\theta} L_{total}$ represents the gradient associated with the function of a total loss considering network parameters. After the training of the encoder is accomplished, the possible abnormalities are identified concerning the reconstruction error considering retinal images which have higher possibilities of DR. The empirical formulation of a reconstruction error for X as an input image is represented as follows:

$$E(X) = \|X - \hat{X}\| \quad (8)$$

According to (8), if the error of reconstruction $E(X)$ is found to be higher than a certain cut-off value T then the image under observation is concluded to be possessing abnormalities. This could further have possibilities of positive DR. The determination of cut-off T is carried out on various experiments on the validation set. The modelling of reconstruction error distribution is carried out by the Gaussian mixture model which is utilized for computing the probabilities of all errors during the reconstruction process i.e. $E(X)$. The idea is towards classifying the normal image from DR. The mathematical representation of the probability of error in image reconstruction is as follows:

$$Prob = \sum_{k=1}^K \alpha_k \cdot gd(s) \quad (9)$$

In (9), the computation of $Prob$ (probability of reconstruction error) is carried out considering α as a mixing coefficient and gd as Gaussian Distribution concerning E as error, mean and standard deviation. The images are further subjected to classification based on the probabilities of their reconstruction error that belongs to a class of DR as follows,

$$C_{pred} = \arg_{\max} Prob(E|C) \quad (10)$$

In (10), the computation of class of prediction C_{pred} is carried out considering a maximum number of arguments associated with computed probability $Prob$ in the previous step concerning error E and class C . By performing this last step, the objective towards the detection and classification of DR is accomplished. The next section outlines the study outcomes.

4. RESULT

The implementation of the proposed study is carried out considering standard retinal image datasets. The study uses DIARETDB1, which consists of 89 images [29], and EyePACS, which comprises 35,000 images [30]. Table 1 showcases the hyperparameters used for the analysis considering standard performance metrics of accuracy, sensitivity, specificity, and F1-Score. Further, the assessment is carried out by comparing the proposed model *Prop*, with CNN, SVM, DBN, and ResNet-50, which are found to be frequently adopted towards investigating DR.

Table 1. Hyperparameters and their adopted values

Hyperparameter	Value/Setting
Autoencoder architecture	Convolutional layers with 32, 64, 128, 256 filters, latent space dimension of 512
Activation function	ReLU (intermediate), Sigmoid (output layer)
Latent space dimension	512
Optimizer	Adam, Learning rate = 0.001, $\beta_1=0.9$, $\beta_2=0.999$, $\epsilon=10^{-7}$
Loss function	Mean Squared Error (MSE), L2 Regularization
Batch size	32
Epochs	50 (with early stopping)
Dropout rate	0.3 (after convolutional layers)
Weight initialization	Xavier Initialization
GMM components	2 (Healthy, DR)
GMM covariance type	Full covariance matrix
GMM max iterations	1000
Train-Test Split	70%-15%-15%

Table 2 and Figures 2-5 showcase the actual study outcome to show that *Prop* excels significantly better performance in contrast to frequently existing learning models towards detection and classification of DR. An interesting fact to observe in this evaluation score exhibited in Table 2 is that it offers nearly similar performance concerning both the datasets and hence its consistency can be eventually ascertained over adopted performance metrics.

Table 2. Comprehensive study outcomes

Model	Dataset	Accuracy (%)	Sensitivity (%)	Specificity (%)	F1-Score
Prop	DIARETDB1	96.5	94.2	98.3	0.96
Prop	EyePACS	95.1	92.8	97.5	0.94
CNN	DIARETDB1	92.1	88.3	95.2	0.90
CNN	EyePACS	93.6	89.5	96.3	0.91
SVM	DIARETDB1	85.4	83.2	89.6	0.84
SVM	EyePACS	87.3	85.1	90.8	0.86
DBN	DIARETDB1	90.2	86.5	93.5	0.88
DBN	EyePACS	91.3	87.9	94.8	0.89
ResNet-50	DIARETDB1	93.4	89.8	95.8	0.91
ResNet-50	EyePACS	94.3	90.2	96.1	0.89

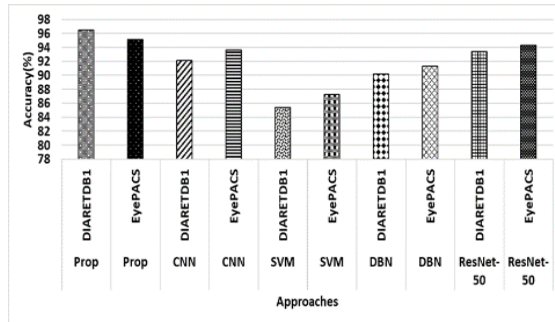


Figure 2. Assessment of accuracy

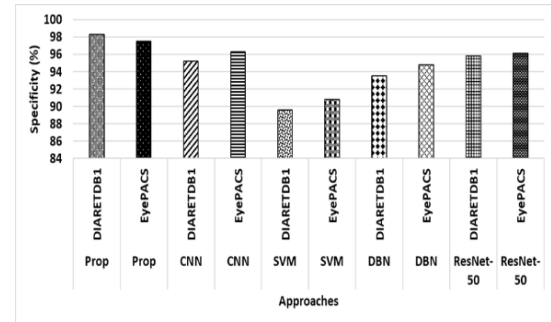


Figure 3. Assessment of specificity

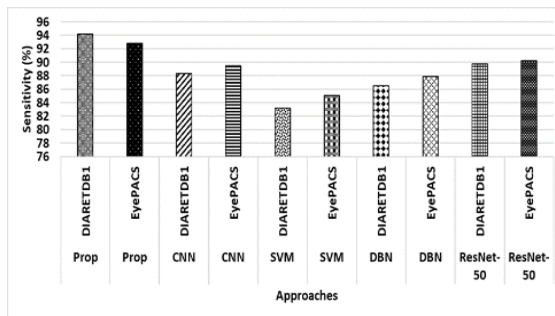


Figure 4. Assessment of sensitivity

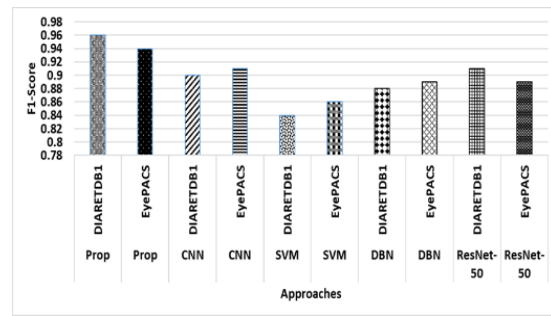


Figure 5. Assessment of F1-score

4.1. Ablation study

Table 3 presents the results of the ablation study, where various components of the model were altered to determine their influence on the overall performance. Each experiment tested the model's performance in terms of accuracy, sensitivity, specificity, and F1-Score using the DIARETDB1 dataset. Table 3 showcases the outcome of the ablation study. With 96.5% accuracy, 94.2% sensitivity, and 98.3% specificity, the full model (Prop) performs best across all measures, demonstrating the value of combining autoencoders, GMM, and other methods. Accuracy and sensitivity significantly decrease when the autoencoder is removed, suggesting that autoencoders are essential for feature extraction in DR detection.

Table 3. Ablation study results on DIARETDB1 dataset

Experiment	Accuracy (%)	Sensitivity (%)	Specificity (%)	F1-Score
Full model (Prop)	96.5	94.2	98.3	0.96
Without autoencoder	92.3	89.1	94.7	0.90
Without GMM	93.8	91.4	96.1	0.92
Without data augmentation	94.1	91.7	96.8	0.93
Without RoI segmentation	95.2	92.8	97.9	0.94
Without regularization (L2)	93.7	90.3	96.4	0.91

Performance is moderately reduced when GMM is disabled, demonstrating the significance of reconstruction error modelling for differentiating between normal and DR pictures. Removing ROI segmentation or data augmentation weakens the model, demonstrating that these methods improve the model's capacity for generalization. Despite having a negligible impact, L2 regularization helps to improve model generalization and lessen overfitting.

4.2. Discussion

The key findings are summarized as follows: we developed a new deep-learning model for the early identification of DR by combining Gaussian mixture models for anomaly detection with CAE. Across several datasets, including DIARETDB1 and EyePACS, the suggested model, known as Prop, outperformed more established models, including CNN, SVM, DBN, and ResNet-50. On the DIARETDB1 dataset, the Prop model demonstrated a remarkable 96.5% accuracy and 94.2% sensitivity, demonstrating its efficacy in identifying DR at an early stage. These findings demonstrate the model's resilience and its capacity to achieve high specificity (98.3%) while reducing false positives.

The Prop model continuously surpassed earlier research, including those conducted with CNN or ResNet-50, especially in terms of sensitivity, which is essential for identifying DR in its early, most curable phases. This was further corroborated by the ablation investigation, which demonstrated that every element—including data augmentation, ROI segmentation, and GMM integration—made a substantial contribution to the model's performance. High classification accuracy and reliable generalization across a variety of datasets were made possible by the combination of autoencoders for feature extraction and GMM for anomaly detection. In summary, our study has shown that a hybrid autoencoder-GMM technique, which offers a balance of high accuracy, sensitivity, and computational economy, is a potential tool for early-stage DR identification. The suggested system offers a strong basis for upcoming developments in automated retinal screening and, eventually, improving patient outcomes by identifying diabetic problems early.

5. CONCLUSION

The current paper has presented a novel deep-learning model using an autoencoder to detect DR in its early stages. By creating a unique autoencoder-based model and integrating it with Gaussian mixture models, this study effectively tackles the main obstacles in the early diagnosis of DR. This greatly improves the sensitivity and specificity of DR detection in its early stages. We show that our method not only achieves high classification accuracy but also guarantees strong generalization by validating the proposed model on several benchmark datasets, achieving the goal of offering a practical, scalable, and interpretable solution for DR diagnosis in real-world clinical settings. The primary novelty and contribution of the study are as follows: i) the issues of early-stage detection identified in related work have been addressed by the proposed hybridization of GMM and Autoencoder by using reconstruction errors for determining granular anomalies associated with retinal images. This directly improves the sensitivity of the system towards early detection of DR. ii) The issue of inadequate generalization over diverse data is addressed in the current work by considering two different datasets of EyePACS and DIARETDB1 for enhanced applicability and improved generalization to near-practical clinical settings. iii) the issues of the Complexity of the Learning Approach are addressed by using an autoencoder for anomaly detection with reduced computational burden and more efficient than ResNet-50, a deeper network. iv) the issues of Sub-optimal interpretability of the deep learning model are addressed by joint integration of GMM with reconstruction error for better interpretability.

Future work will be carried out towards benchmarking the proposed system with other associated ocular diseases like glaucoma. A novel approach that can distinguish DR from glaucomic patients in its early stage will be developed harnessing a more innovative AI-based approach.

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Kavita V. Horadi		✓				✓		✓	✓	✓	✓	✓		
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C : Conceptualization	I : Investigation	Vi : Visualization
M : Methodology	R : Resources	Su : Supervision
So : Software	D : Data Curation	P : Project administration
Va : Validation	O : Writing - Original Draft	Fu : Funding acquisition
Fo : Formal analysis	E : Writing - Review & Editing	

CONFLICT OF INTEREST STATEMENT

Authors state no conflict of interest.

DATA AVAILABILITY

The data that support the findings of this study are available from the corresponding author upon reasonable request.




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