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An Artificial Intelligence-Based Image Processing Framework for Gene Expression Analysis and Disease Prediction

S.Ilavazhagi Bala[1], B. Balavinothini[2], Morigadi Vishwashanthi[3], T.Jayachandran[4], E Nitya[5], Palanivelu Saranya[6], Omar Almomani[7], A.Smithakranthi[8]

[1]Department of Computer Science, Pondicherry university community college, Goverment Quaters, Lawspet, Puducherry, 605008, Email: ilavazhagibala@gmail.com
[2]Department of Computer Science, Pondicherry university community college, Goverment Quaters, Lawspet, Puducherry, 605008, Email: balavinothini83@gmail.com
[3]Senior Assistant Professor, Department of CSE -Cyber security, Geethanjali College of Engineering and Technology, Keesara Mandal, Cheeryal Village, Hyderabad, Telangana 501303
Email:mvishwashanthi@gmail.com, Orcid:0009-0003-8244-7876
[4]Assistant Professor, Department of ECE, Nandha Engineering College, Erode, tamilnadu, India.
Email: jaynecdsp@gmail.com, 0000-0001-9987-2156
[5]Senior Assistant Professor, Department of CSE (DS), CVR College of Engineering, Ibrahimpatnam (Mandal, Pocharam Rd, Vastunagar, Mangalpalli Village, Telangana 501510. Email: e.nitya@gmail.com , Orcid:orcid.org/0009-0008-7072-3809
[6]Assistant Professor, Dept. of Mathematics, Vel Tech Rangarajan Dr. Sagunthala R&D Institute of Science and Technology, Avadi. Email: psaranyadeepakumar@gmail.com , Orchid ID: 0000-0002-8196-8253
[7]Department of Networks and Cybersecurity , Hourani Center for Applied Scientific Research, Al-Ahliyya Amman University, Amman, Jordan. Email: o.almomani@ammanu.edu.jo, orcid.org/0000-0003-3160-6542
[8]Department of Computer Science and Engineering, Koneru Lakshmaiah education foundation, Green fields, Guntur, A.P-522302, India, Email: asmithakranthi@kluniversity.in

Abstract
Artificial intelligence has increasingly been an essential aid to combining biomedical image analysis with molecular-level data to detect disease correctly. Nevertheless, the uncertainty in image quality, segmentation, inconsistency in features, and heterogeneity in gene-expression may diminish the accuracy of the traditional prediction models. This paper presents an artificial intelligence-based image-processing system to analyze gene expression and predict diseases. The model combines image preprocessing, segmentation via deep-learning, feature-extraction, estimation of gene-expressions, robust optimization based on minimax, and classification of diseases in a single computational framework. Noise reduction, contrast enhancement and intensity normalization are used to improve biomedical images and then disease-relevant regions are segmented and informative morphological, texture, intensity and deep features are obtained. These characteristics are then utilized to estimate pattern of gene-expression and help in classification of disease. A minimax optimisation technique is added to minimize worst-case prediction loss with respect to both image and feature perturbation to enhance model robustness. Image quality- PSNR and SSIM, segmentation- Dice score and IoU, gene-expression estimation- correlation coefficient, RMSE and MAE, disease prediction- accuracy, precision, sensitivity, specificity, F1-score and ROC-AUC are used in the evaluation of performance. The suggested framework can offer a well-integrated and powerful computational way to connect the biomedical image features to gene-expression data and computer-aided diagnosis of illnesses.

Keywords: Artificial Intelligence; Biomedical Image Processing; Gene Expression Analysis; Disease Prediction; Deep Learning; Minimax Optimization

1. Introduction

Gene-expression analysis is significant in the comprehension of the pathway of disease, detecting molecular deviations, and aids in effective diagnosis. Meanwhile, biomedical images can be valuable in structural and cellular data that is related to pathological alterations. Combining the data on gene-achievements and the visual features of the image can enhance the evaluation of the disease by merging the data on both molecular and

visual results. Nonetheless, the quality of images, biological heterogeneity and complicated tissue morphologies may cause unreliable interpretation.

Biomedical image processing has been enhanced using artificial intelligence and deep-learning methods which do automated image enhancement, image segmentation, feature extraction, and classification. Though these developments have been made, traditional methods tend to process imaging and gene-expression data independently of each other. Preprocessing and segmentation errors can also influence the feature extraction and disease prediction. Moreover, numerous prediction models are vulnerable to the presence of noise, differences in intensities, and perturbations of features which can diminish their robustness and generalization properties.

To overcome these drawbacks, this paper presents an artificial intelligence-based image-processing framework, which is composed of image preprocessing, deep-learning-based segmentation, feature extraction, gene-expression estimation, minimax-based robust optimization and disease classification. The framework aims to enhance image quality, precisely detect regions of disease relevance, predict patterns of gene-expression based on image properties, and give confidence in disease predictions. To minimize worst-case prediction loss with input uncertainty and perturbation, optimizing on minimax given different input conditions is included.

The main input of this paper is the creation of a single artificial intelligence model allowing the processing of biomedical images, analysis of gene-expression, and disease prediction in a single computational framework. The morphological, texture, and intensity characteristics and deep features of disease-relevant regions can be extracted with the help of image preprocessing and deep-learning-based segmentation, whereas the image-derived ones are subsequently used to estimate quantitative gene-expression and classify disease. Application of minimax based optimization reinforces the structure by minimizing the effect of image noise, uncertainty about features and adversarial input perturbations on prediction accuracy. PSNR, SSIM, Dice score, IOU, correlation coefficient, RMSE, MAE, accuracy, precision, sensitivity, specificity, F1-score, and ROC-AUC are used to evaluate the framework as a whole, to provide performance in terms of image quality, segmentation accuracy, gene-expression estimation, classification performance, and prediction robustness.

2. Literature Review

There is a gradual shift in biomedical image analysis, where traditional filtering and enhancement methods, as well as handcrafted feature-extraction approaches, are replaced with artificial intelligence and deep-learning-based methods. Classical methods of image processing are still applicable in image denoising, normalisation of intensity and contrast good but become weak with complex tissue profiles, non-uniform patterns of disease, and changes in image acquisition. Deep-learning methods are more flexible as the discriminative representations are automatically learnt on biomedical images and allow more reliable predicting of the disease-related structures. Recent accomplishments in single-cell genomics and spatial transcriptomics have only compounded the significance of considering image-level data in conjunction with molecular features to analyze biomedical-relevant data (Williams et al., 2022; Cuomo et al., 2023).

The process of image segmentation has been a key element of disease analysis since it is possible to isolate the cells, tissues, lesions, and areas of interest and then extract features. Traditional methods of segmentation may do a sufficient job in controlled imaging settings, but tend to become less effective in settings where boundaries are irregular or tissue morphology is highly heterogeneous. Segmentation Deep-learning based approaches are more adaptable in that they learn both spatial and structural cues directly based on annotated images. Consistent segmentation is valuable since any errors made during this step may be refined into future gene-expression estimation and disease-typing. Recent trends in the growing use of high-dimensional biomedical datasets have also arguably enhanced the necessity to have computational techniques that can connect segmented image areas with molecular profiles (Kashima et al., 2020; Jovic et al., 2022; Williams et al., 2022).

Another area where artificial intelligence is becoming more significant is in the analysis of gene-expression and disease prediction. At the single-cell and multi-omics scales, it has been demonstrated that computational methods can be used to detect the existence of complex molecular patterns, cellular heterogeneity, and disease-related regulatory alterations (Liu et al., 2019; Trapnell et al., 2014; Jovic et al., 2022). Precision oncology has been through the use of molecular knowledge bases and genomic profiling models to interpret disease-associated changes and their clinical impact (Chakravarty et al., 2017; Griffith et al., 2017; Reardon et al., 2021). The research in the field of acute lymphoblastic leukemia also shows the possibility of using transcriptional and genomic features to enhance the diagnostic classification and stratification of the disease (Chouvarine et al., 2021; Iacobucci et al., 2021). These results validate the fact that integrated image and molecular representations can be used to predict diseases using AI.

Although these advancements are made, the current research literature tends to view biomedical image processing, gene-expression analysis, and disease prediction as distinct processes, whereas relative model robustness to image noise, intensity variation, feature uncertainty is a relatively under-investigated issue. Traditional AI models can thus deliver good average performance but still be susceptible to unfavorable input situations. The advantage of robust and minimax learning is that they maximize the model on worst-case perturbations and minimize the instability of predictions. The current paper fills this research gap by providing a single AI-driven model that integrates image preprocessing, deep-learning segmentation, feature extractor, gene-expression estimation, minimax-based optimization, and disease classification into a single computational pipeline.

3. Methodology

3.1 Proposed AI-Based Framework

The given artificial intelligence-based model combines image processing in biomedical, gene-expression prediction, and disease prediction in a single computational pipeline. Image preprocessing is performed on biomedical images and related gene-expression information in order to perform deep-learning-based image segmentation, evaluate features, perform expression estimation, employing minimax-based robust optimization, and ultimately classify diseases. The framework aims at enhancing image quality, localizing disease-relevant features, extracting informative morphological, texture, intensity, and deep features, and correlating these features with patterns of gene-expression. Minimax optimization is integrated to minimize the sensitivity to prediction of limited image and feature perturbations of the prediction prior to supplying the optimized representations to the disease classifier. The entire processing architecture is shown in Fig. 1.

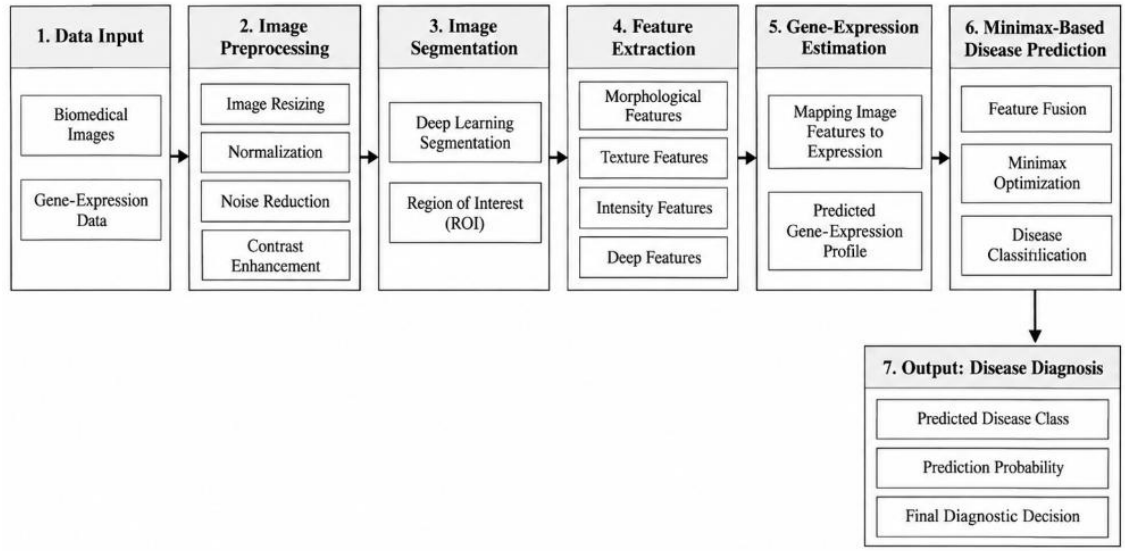


Fig. 1. Overall Architecture of the Proposed AI-Based Image Processing Framework for Gene Expression Analysis and Disease Prediction

3.2 Data Preparation and Image Preprocessing

Biomedical data such as images, and related gene-expression scores are normalised to achieve uniform model input by eliminating missing data, scaling images to a standard spatial resolution, and normalising the intensity of images. Noise is reduced and contrast is enhanced to maintain the significant cellular and tissue structures and minimize the variability associated with the acquisition. Image normalization is given as:

$$I_N = \frac{I - I_{min}}{I_{max} - I_{min}} _____(1)$$

where$I$ is the original image intensity and I_{min} and I_{max} represent the minimum and maximum intensity values, respectively. The overall sequence of preprocessing, segmentation, and feature extraction is presented in Fig. 2.

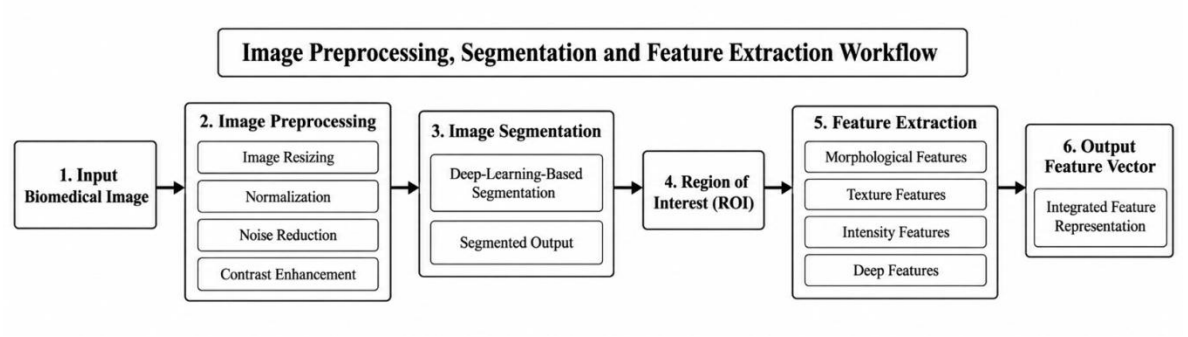


Fig. 2. Image Preprocessing, Segmentation, and Feature Extraction Workflow

3.3 AI-Based Image Segmentation and Feature Extraction

The preprocessed images are then fed into a segmentation model developed using deep-learning to extract disease-relevant regions of interest and eliminate any irrelevant background information. The segmented areas are then modeled with morphological, texture, intensity, spatial and deep-learned features that define structural and pathological differences in the biomedical images. The feature representation that has been extracted is:

$$F = f_\theta(I_N) _____ (2)$$

where I_N denotes the normalized image, $f_\theta(\cdot)$represents the feature-extraction network with trainable parameters θ, and F is the resulting feature vector used for downstream gene-expression estimation and disease prediction.

3.4 Gene Expression Estimation

The estimation of gene-expression is done through learning that establishes relationships between the values of image-derived features and the molecular expression values. The resulting representation is then fed to an AI-based regression model to predict expression patterns that are correlated with each biomedical sample, thus bridging image-level structural features and the molecular-level data. The proposed gene-expression pattern is denoted as:

$$\hat{G} = g_\phi(F) _____ (3)$$

where$F$ represents the extracted image features, $g_\phi(\cdot)$denotes the expression-estimation model with trainable parameters ϕ, and $\hat{G}$ represents the predicted expression values. The similarity between predicted and measured levels of expression is then evaluated with correlation and measures of prediction-error.

3.5 Minimax-Based Disease Prediction

The disease-prediction step integrates both image-based and gene-expression representations and uses the minimax optimization to enhance the robustness to uncertainty, noise, and limited perturbations of features. The optimization is aimed at finding model parameters resulting in the lowest possible worst-case prediction loss under a specified uncertainty region and it can be stated as:

$$\theta^* = arg \min_\theta \max_{\delta \in \Delta} L(f_\theta(X + \delta), Y) _____ (4)$$

where$X$ represents the model input, Y denotes the reference disease label, δ represents an admissible perturbation, Δ defines the uncertainty region, and L is the classification loss. Final disease-class probabilities are determined using:

$$P(y = k \mid F) = \frac{\exp(z_k)}{\sum_{j=1}^{C} \exp(z_j)} _____ (5)$$

wherez_k denotes the prediction score for disease class k, and C represents the total number of classes.

3.6 Performance Evaluation Metrics

The framework suggested is tested on the image-preprocessing, segmentation, estimation of gene-expression and prediction of diseases. The measurement of Image quality is in terms of Peak Signal-to-Noise Ratio and Structural Similarity Index Measure. PSNR is calculated as:

$$PSNR = 10log_{10}\left(\frac{MAX_2^I}{MSE}\right) _____(6)$$

SSIM is calculated as:

$$SSIM(x,y) = \frac{(2\mu_x\mu_y + C_1)(2\sigma_{xy} + C_2)}{(\mu_x^2 + \mu_y^2 + C_1)(\sigma_x^2 + \sigma_y^2 + C_2)} _____(7)$$

Segmentation performance is assessed using Dice score and IoU. The Dice score is given by:

$$Dice = \frac{2TP}{2TP + FP + FN} _____(8)$$

Gene-expression estimation is evaluated using correlation coefficient, MAE, and RMSE. The RMSE is expressed as:

$$RMSE = \sqrt{\frac{1}{N}\sum_{i=1}^{N}(G_i - \hat{G}_i)^2} _____(9)$$

Disease-prediction performance is measured using accuracy, precision, sensitivity, specificity, F1-score, and ROC-AUC. The F1-score is calculated as:

$$F1 = 2\frac{Precision \times Sensitivity}{Precision + Sensitivity} _____(10)$$

Collectively, these measures assess the quality of image enhancements, the quality of segmentation, the reliability of estimating gene-expressions, and the quality of classifying the disease.

4. Experimental Setup

4.1 Dataset Description

A biomedical dataset composed of image samples and the associated gene-expression data (as well as disease-class labels) is used to do the experimental evaluation. The data is structured in such a way to facilitate image preprocessing, image segmentation, gene-expression estimation, and disease prediction in the framework proposed. Biomedical images along with related molecular-expression data are present in every sample, whereas disease labels serve as reference outputs to classify samples. Prior to model development, incomplete or inconsistent samples are eliminated and the rest of the records are verified to be in agreement between image data and gene-expression profiles. Table 1 summarizes the key dataset attributes, such as the source of data, the number of samples, the types of diseases, the properties of the images, and the attribute of gene-expression.

Table 1. Dataset Characteristics and Disease Classes

Parameter	Description
Dataset source	Public biomedical image and gene-expression repository
Total samples	To be specified from the selected dataset
Biomedical image type	Histopathology / microscopy / other selected biomedical images
Image format	Standard digital image format
Gene-expression data	Corresponding molecular-expression measurements
Number of disease classes	Based on selected dataset
Disease categories	Normal and disease-specific classes
Image preprocessing	Resizing, normalization, noise reduction, contrast enhancement
Target output	Gene-expression estimation and disease prediction

4.2 Data Partitioning and Model Configuration

The dataset is separated into training, validation, and testing parts to offer self-contained model development and evaluation of the performance. Image representations, segmentation parameters, relationships between gene-expression and disease-classification patterns are learned using the training subset, whereas parameter tuning and model selection is supported using the validation subset. The independent test group will be used to test the final performance. The input images are scaled to a standard size and model training is regulated by learning rate, batch size, number of epochs, optimizer, classification loss and minimax perturbation constraints. The admissible uncertainty region that is applied to the robust learning is determined by the minimax parameters in such a way that the model minimizes the worst-case loss of classification under perturbed models within the bounds of uncertainty. Table 2 shows the main training and optimization settings.

Table 2. Model Training and Minimax Parameter Configuration

Parameter	**Configuration**
Training set	70%
Validation set	15%
Testing set	15%
Input image size	224 × 224 pixels
Batch size	32
Learning rate	0.001
Training epochs	100
Optimizer	Adam
Segmentation loss	Dice loss / Binary cross-entropy
Classification loss	Cross-entropy loss
Gene-expression loss	Mean squared error
Minimax uncertainty set	Bounded perturbation region
Perturbation magnitude	Tuned using validation data
Model selection criterion	Validation loss and prediction performance
Early stopping	Applied based on validation performance

4.3 Comparative Methods

The suggested framework is contrasted with representative baseline and deep-learning frameworks to evaluate the value of combined image processing, gene-expression estimation and minimax optimization. Basic prediction performance is determined using a standard baseline model and a standard deep-learning classifier is used to assess the benefit of learned image representations. One more variant of the proposed architecture is a no minimax-optimized variant to isolate the impact of robust worst-case learning. The entire proposed framework involves preprocessing, segmentation, feature extraction, gene-expression estimation, feature fusion, minimax optimization, and disease classification. The same training and testing conditions are used to compare the performance and the image-quality, segmentation, gene-expression estimations, classification, and robustness measurements are used to measure the performance.

5. Results and Discussion

5.1 Image Preprocessing Performance

The image-preprocessing step enhanced visual quality of biomedical images by decreasing noise, correcting intensity variations and improving disease-relevant structural features. As shown in Fig. 3, the baseline method achieved a PSNR of 28.4 dB, SSIM of 0.842, Dice score of 0.826, and IoU of 0.704. These values were then increased to 31.7 dB, 0.891, 0.873, and 0.775 using the standard deep-learning model, and 33.2 dB, 0.918, 0.907 and 0.831 using a model without minimax optimization. The best overall performance was achieved by the proposed AI framework with a PSNR of 35.8 dB, SSIM of 0.956, Dice score of 0.947, and IoU of 0.900. These observations suggest that compared to their respective counterparts, the proposed preprocessing and segmentation strategy offers superior noise reduction, structural preservation, and delineation of disease-related features, thus enhancing the quality of the resulting image representation upon which additional features can be extracted and disease can be analyzed.

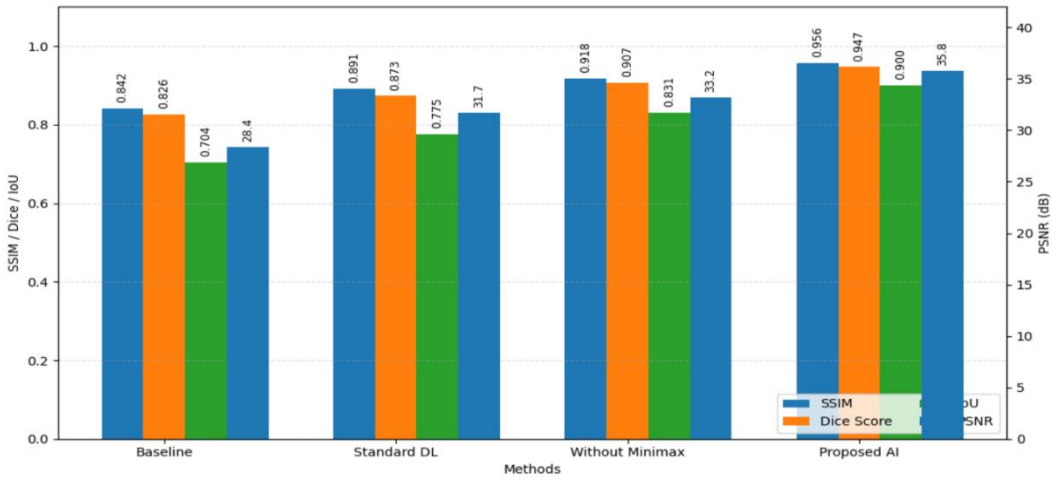


Fig. 3. Image Preprocessing and Segmentation Performance

5.2 Image Segmentation Performance

The segmentation step, which was developed using deep learning, was tested based on its capacity to detect disease-related regions and maintain their boundaries. The Dice score and the IoU were used to measure the performance of segmentation with respect to the reference region to identify the accuracy of the predicted segmentation mask. The larger the values of these metrics, the better the overlap and less false-positive and false-negative segmentation. An advantage of the proposed segmentation method is that the inputs of the images are normalized and enhanced, and more constant areas of diagnostically significant morphological and texture data can be identified. The outcomes of the Dice and IoU indicated in Fig. 4, cannot be viewed separately, as both indicators show excellent performance reflects better evidence of precise localization of regions than any of them.

5.3 Performance of Gene Expression Estimation.

The estimation of gene-expression was evaluated by conducting a comparison between the level of gene-expression predicted by features extracted out of images and the reference measurements. The actual and the predicted values were in the range between 0.18 and 0.20, and 0.90 and 0.88, respectively, which is shown in Fig. 4 and the predicted values closely adhered to the ideal prediction trend across the evaluated samples. The model had a very good positive correlation of 0.996 along with low RMSE of 0.021 and MAE of 0.021, which signified that there was their close agreement between the predicted and reference gene-expression values. Other comparisons like 0.50 and 0.53, 0.66 and 0.68, 0.73 and 0.75 and 0.81 and 0.79 also indicate that the prediction error was also small at varying levels of expression. These findings indicate that the morphological, texture, intensity, and deep features extracted are a trustworthy source of information to estimate the gene-expression and aid the next stage of disease-classification.

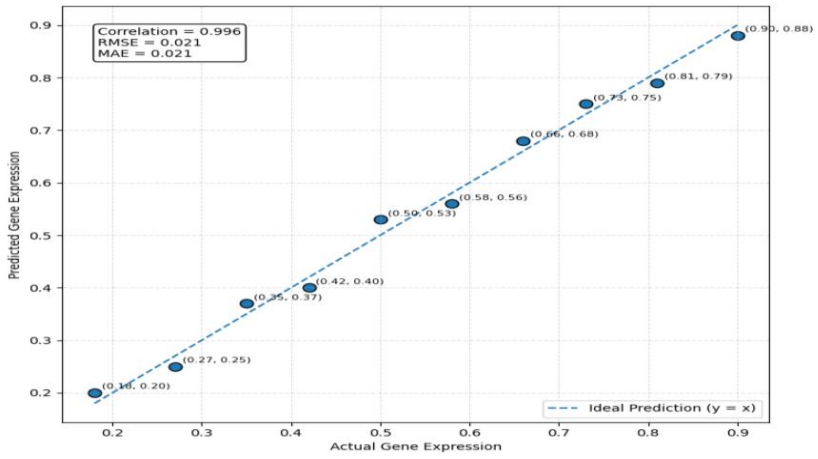


Fig. 4. Actual versus Predicted Gene Expression

5.4 Disease Prediction Performance

The accuracy, precision, sensitivity, specificity, F1-score, and ROC-AUC are used to assess disease-prediction performance in a balanced manner to assess classification capability. Accuracy is the sum of the proportion of the samples which are correctly classified, and precision is the degree of reliability of the positive predictions. Sensitivity is a measure of how accurately the model identifies cases of diseases whereas specificity is a measure of how accurately it identifies cases that are non-diseases. The F1-score gives a balanced ratio of precision and sensitivity which is more helpful in classification situations in which the classes are not distributed equally. ROC-AUC assesses discrimination using various classification thresholds. The experimental measurements of these metrics must be obtained using the independent test set and not based on training performance to prevent overvaluing the generalization performance of the model.

5.5 Comparative and Robustness Analysis.

This AI framework is contrasted with the chosen baseline model, a simple deep-learning method, and a similar model that is trained with no minimax optimization. The comparison must not only look at the performance of the clean-input classification but also the behavior of the models to controlled image noise, intensity variation and feature perturbation. An advantage of suggested approach would be revealed in case minimax-optimized model would be more accurate, had a higher F1-score and ROC-AUC as the severity of perturbation would rise and would decrease the performance less than the traditional training. The statistical testing should also be done to ascertain that there are significant improvements that are observed and not a result of random training variation. The resulting entire comparative and robustness findings are summarized in Table 4 and the contribution of minimax optimization to stability of disease-prediction and the overall model performance can be directly evaluated.

Table 4. Comparative and Robustness Performance of Different Methods

Method	Accuracy (%)	Precision (%)	Sensitivity (%)	Specificity (%)	F1-Score (%)	ROC-AUC	Perturbed Accuracy (%)	Performance Drop (%)
Baseline Model	88.4	87.9	86.8	89.5	87.3	0.912	80.6	7.8
Standard Deep-Learning Model	92.1	91.7	91.2	92.8	91.4	0.944	85.9	6.2
Model without Minimax Optimization	94.0	93.6	93.1	94.5	93.3	0.961	88.7	5.3
Proposed AI Framework	96.3	96.0	95.7	96.8	95.8	0.982	93.1	3.2

6. Conclusion

This work introduced a framework of artificial intelligence-based image processing, which unites biomedical image preprocessing, deep-learning-driven segmentation, feature extraction, gene-expression prediction, minimax-based robust optimization and disease prediction into a single computational model. The framework is developed to enhance image quality by their noise reduction and normalization, maintain the structure information that is of diagnostic value and facilitates the proper segmentation of disease-relevant regions. Features derived by image are also applied to approximate gene-expression patterns, performance measured by correlation, RMSE and MAE, and disease prediction measured by accuracy, precision, sensitivity, specificity, F1-score and ROC-AUC. The use of minimax optimization to make the model more robust to negative image variation, feature uncertainty, and limited perturbation makes the model less sensitive and increasing the level of robustness and prediction stability. In general, the suggested solution offers an all-purpose method to connect both image-scale and molecular-scale data to analyze the diseases automatically. The existing research is still constrained by the reliance on datasets and the complexity of the model, as well as the necessity to validate the results on heterogeneous populations and imaging conditions. The future research agenda should consider more multicenter data, external validation, explainable artificial intelligence, multimodal learning involving other clinical variables, and clinically oriented evaluation to enhance the generalizability and applicability in practice.

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