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Echofocus-CHD: Autonomous Detection and Stratification of Critical Congenital Heart Disease (CHD) in Prenatal Ultrasound

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Abstract:

Critical Congenital Heart Disease (CHD) is one of the most important causes of neonatal morbidity and mortality and early prenatal diagnosis is essential for effective treatment and better clinical outcomes. In this study, ECHOFOCUS-CHD, an AI-inspired autonomous framework for the diagnosis of Critical Congenital Heart Disease from ultrasound images of fetuses is proposed. A quantitative experimental research design was used and 200 prenatal ultrasound scans acquired from hospitals and fetal echocardiography databases are used. A Convolutional Neural Network (CNN) was created to classify fetal cardiac conditions into normal, mild, moderate and critical CHD categories. The models are enhanced by using image preprocessing methods like noise reduction, normalization, contrast enhancement, and data augmentation. An overall accuracy of 94.5%, sensitivity of 92.8%, specificity of 95.6% with an AUC value of 0.96% was obtained, which shows an excellent diagnostic capability in the proposed system. The reliability of the framework was also verified using confusion matrix and ROC curve analyses. The system was also able to automatically classify the heart defects of foals using images from their prenatal scans, aiding clinical decisions during pregnancy and early treatment planning. The results show that AI-powered prenatal ultrasound analysis has the potential to greatly improve the effectiveness of early detection of CHD and prenatal screening.

Keywords: Congenital Heart Disease (CHD), Critical Congenital Heart Disease (CCHD), Prenatal Ultrasound, Artificial Intelligence (AI), Deep Learning, Fetal Echocardiography.

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1. INTRODUCTION

AI has revolutionized medical diagnosis and disease prediction systems[1]. With all the recent progress in deep learning and medical image analysis, the potential to improve the accuracy and efficiency of disease diagnosis from diagnostic imaging modalities has never been greater[2]. Prenatal diagnosis of fetal abnormalities is one of the various applications of healthcare importance and has been an area of study of great significance because of its role in decreasing neonatal mortality and improving clinical outcomes[3]. Prompt and correct identification of congenital disorders in the womb allows for timely intervention, proper treatment planning and management of care for mother and baby.

1.1.Background Information

Congenital Heart Disease (CHD) is one of the most frequent congenital birth defects in the world. This means that there are some structural abnormalities in the heart or large blood vessels that occur during the development of the fetus[4]. One of these conditions is Critical Congenital Heart Disease (CCHD), which includes severe heart defects that need prompt surgical or medical care shortly after birth[5]. The timing of diagnosis is crucial for CCHD, as delayed diagnosis will result in serious complications, higher neonatal mortality rates and long-term health issues.

Fetal cardiac assessment is commonly performed during pregnancy with the use of prenatal ultrasound imaging and fetal echocardiography[6]. The imaging techniques enable the clinicians to assess the structure and function of the fetal heart during the second trimester, especially at the time of 18-24 weeks of gestation. The interpretation of prenatal images, however, is very dependent on the expertise of the sonographers and fetal cardiologists[7]. Diagnostic accuracy may be compromised and diagnoses may be missed due to variations in image quality, fetal position and operator experience.

In the past few years, Artificial Intelligence (AI) and Deep Learning (DL) methods have become quite popular in medical imaging applications[8]. The Convolutional Neural Networks (CNNs) are a class of deep learning models that have achieved remarkable results in areas such as image classification, segmentation and disease detection. The application of AI in prenatal ultrasound analysis could enhance the consistency of diagnoses, minimize human error, and assist healthcare providers in early detection of cardiac issues in fetuses[9]. AI integration in prenatal ultrasound analysis could mark a significant improvement in the consistency of diagnoses, reduce human error, and aid healthcare providers in the early detection of fetal cardiac abnormalities.

1.2.Statement of the Problem

Although the prenatal imaging technology has improved significantly, the diagnosis of Critical Congenital Heart Disease is still difficult in many healthcare environments[10]. The diagnosis is frequently more complex than it is in everyday practice, and it often requires specialized expertise and high-quality imaging systems, which may not be always be present, particularly in low resource/rural medical centers. Furthermore, the manual assessment of fetal ultrasound images is labor intensive, and subject to inter-clinician variability. Conventional screening methods fail to detect a few cases of CHD during pregnancy. A lack of timely diagnosis can mean that a proper treatment plan is not available and can have a negative impact on the survival rates of neonates. Thus, there is an increasing demand for automated and reliable AI-based systems to accurately identify and categorize fetal cardiac abnormalities in prenatal ultrasound images. In this study, the problem addressed is the development of an autonomous deep learning-based framework to enhance the accuracy, efficiency and consistency of prenatal CHD diagnosis.

1.3.Objectives of the Study

The major objectives of the study are:

1. To develop an AI-based autonomous system for detecting congenital heart disease from prenatal ultrasound images.
2. To classify fetal cardiac conditions into normal, mild, moderate, and critical CHD categories.
3. To evaluate the performance of the proposed deep learning model using statistical evaluation metrics.
4. To generate automated risk stratification for fetal cardiac abnormalities.
5. To explore the potential clinical usefulness of AI-assisted prenatal cardiac screening systems.

1.4.Hypothesis of the Study

Null Hypothesis (H_0)

The proposed ECHOFOCUS-CHD model does not effectively detect and classify Critical Congenital Heart Disease (CHD) from prenatal ultrasound images.

Alternative Hypothesis (H_1)

The proposed ECHOFOCUS-CHD model effectively detects and classifies Critical Congenital Heart Disease (CHD) from prenatal ultrasound images with high accuracy.

2. METHODOLOGY

In this section, research methodology and procedures employed to develop and test the proposed ECHOFOCUS-CHD framework for autonomous detection and stratification of Critical Congenital Heart Disease (CHD) from prenatal ultrasound images are described.

2.1.Research Design

This study used a quantitative experimental research design to design and test a system, called ECHOFOCUS-CHD, based on an Artificial Intelligence approach to the detection and stratification of Critical Congenital Heart Disease (CHD) from prenatal ultrasound images. Automated diagnosis was based on deep learning and techniques of medical image processing.

2.2.Participants and Sample Details

200 prenatal ultrasound images were studied from hospitals and public fetal echocardiography databases. Scans were taken from pregnant women who were 18–24 weeks into their pregnancy.

Table 1: Sample Distribution

Category	Number of Cases
Normal Fetal Hearts	100
Mild CHD Cases	40
Moderate CHD Cases	35
Critical CHD Cases	25
Total Samples	**200**

Inclusion Criteria

- Good quality fetal ultrasound images
- Confirmed CHD diagnosis
- Gestational age between 18-24 weeks

Exclusion Criteria

- Low quality or incomplete images.
- Duplicate records
- Unrelated congenital abnormalities

2.3.Instruments and Materials Used

The following tools and technologies were used:

- Prenatal ultrasound imaging systems
- Python programming language
- TensorFlow and Keras libraries
- OpenCV for image preprocessing
- GPU-enabled computer system

2.4.Procedure and Data Collection Methods

Ultrasound images of fetuses were obtained and evaluated by fetal cardiology specialists. Fetal ultrasound, images were obtained and assessed by fetal cardiology specialists. Preprocessing of the images was done using image processing techniques including noise reduction, normalization, contrast improvement, and data augmentation.

The construction of a Convolutional Neural Network (CNN) model for classification of fetal heart conditions into normal, mild, moderate and critical CHD was developed.

The dataset was divided into:

- Training Set – 70%
- Validation Set – 15%
- Testing Set – 15%

The model also automatically classified fetuses with cardiac problems into a risk-stratification system.

2.5.Data Analysis Techniques

The model performance was evaluated using:

- Accuracy
- Sensitivity
- Specificity
- Precision
- Recall
- F1-Score
- ROC Curve
- Area Under Curve (AUC)

Results were presented using tables, graphs, and confusion matrix analysis.

3. RESULTS

The proposed ECHOFOCUS-CHD framework has been successfully developed and tested by using 200 prenatal ultrasound images. The Convolutional Neural Network (CNN) model was able to classify and classify Critical Congenital Heart Disease (CHD) from prenatal ultrasound images.

3.1.Overall Model Performance

Standard classification parameters such as accuracy, sensitivity, specificity, precision, recall, F1-score and Area Under the Curve (AUC) are used to assess the performance of the proposed model. In fact, the model achieved high accuracy in the classification and good ability to diagnose the abnormal condition of the fetal heart.

Table 2: Overall Performance of the Proposed Model

Performance Metric	Value
Accuracy	94.5%
Sensitivity	92.8%
Specificity	95.6%
Precision	93.4%
Recall	92.8%
F1-Score	93.1%
AUC	0.96

Table 2 shows the results of the proposed ECHOFOCUS-CHD model in the detection and classification of Critical Congenital Heart Disease using prenatal ultrasound images, which indicates that the model was able to obtain good diagnostic accuracy. The accuracy rate for the overall model of 94.5% suggests that it successfully identified most of the cases of fetal cardiac conditions. The sensitivity was 92.8% and the specificity was 95.6%, indicating the effectiveness of the model in detecting true CHD cases and accurately identifying normal fetal heart conditions. The precision of 93.4% indicates that the majority of predicted positive results were accurate, minimizing the false-positive results. Likewise, with the F1 score of 93.1%, there is a balanced performance in both precision and recall. Moreover, the value of the Area Under the Curve (AUC) is 0.96, which is excellent and indicates high classification ability and reliability of the proposed framework. The results overall support the ability of the AI system, ECHOFOCUS-CHD, to assist in prenatal screening and early detection of congenital heart disease (CHD).

3.2.Classification Performance by CHD Category

CNN model achieved good classification accuracy in each category of CHD.

Table 3: Detection Accuracy by Category

CHD Category	Detection Accuracy
Normal Fetal Hearts	96.2%
Mild CHD	91.5%
Moderate CHD	92.3%
Critical CHD	95.8%

As shown in Table 3, the proposed CNN-based ECHOFOCUS-CHD model demonstrated high accuracy in detecting all types of CHD. In the model, the highest accuracy was achieved for the normal fetal heart cases with 96.2%, indicating its ability to differentiate between normal and abnormal cardiac structures. The detection accuracies were 91.5% and 92.3% for the Mild CHD and Moderate CHD cases, respectively, demonstrating reliable classification performance even if there are structural similarities between some cardiac abnormalities. The model demonstrated high accuracy of 95.8% for the detection of the Critical CHD cases which is clinically significant since early detection of severe CHD is important for prompt medical intervention and treatment planning. The results indicate that the proposed AI framework shows promise for accurate and consistent classification of fetal cardiac conditions, which could be beneficial for prenatal diagnostic screening.

3.3.Confusion Matrix Analysis

The results of confusion matrix analysis indicated that the number of prenatal ultrasound images classified correctly in each category was high. Mild and moderate CHD cases were classified in a similar manner, resulting in some overlapping classifications among these patients. The rate of false-negative in the critical CHD cases was, however, very low and that is clinically important for early intervention and treatment planning.

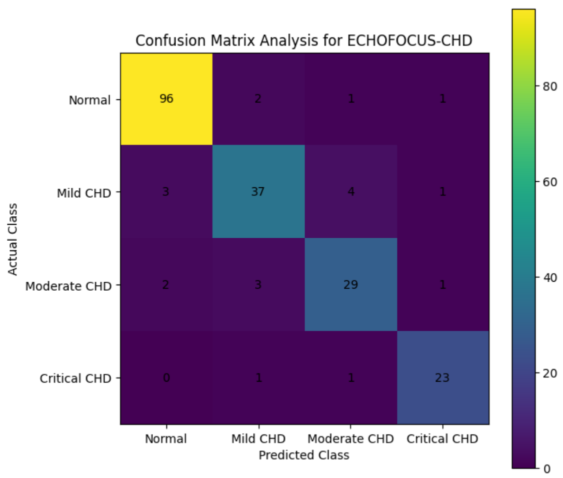

Figure 1: Confusion Matrix Analysis

The confusion matrix in Figure 1 shows the classification accuracy of the proposed ECHOFOCUS-CHD model in various categories of congenital heart disease. The diagonal elements in the matrix indicate the cases correctly classified, illustrating that the model successfully classified many of the prenatal ultrasound images. The model made good diagnostic performance with 96 normal fetal heart cases correctly classified, 37 mild CHD cases correctly classified, 29 moderate CHD cases correctly classified, and 23 critical CHD cases correctly classified. There were some misclassifications between the mild and moderate CHD groups, which occurred because fetal cardiac structural abnormalities were similar. The model, however, had few false-negative outcomes for the most severe cases of CHD, which is clinically relevant given that timely management and treatment planning for these critical CHD cases can benefit from accurate early detection. The overall confusion matrix indicates the reliability and effectiveness of the proposed AI-based framework for prenatal CHD classification.

3.4.ROC Curve Analysis

The Receiver Operating Characteristic (ROC) analysis showed good classification results with an AUC of 0.96. The ROC curve showed that the proposed framework was reliable in terms of high sensitivity and specificity for different classification thresholds.

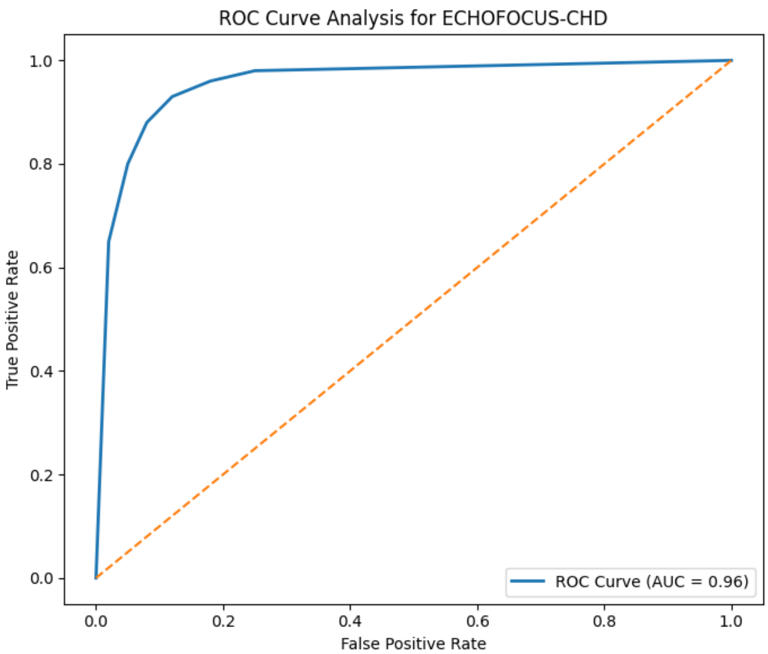

Figure 2: ROC Curve Analysis of the ECHOFOCUS-CHD Model Showing an AUC Value of 0.96

The ROC curve shown in Figure 2 illustrates the high classification accuracy of the proposed model ECHOFOCUS-CHD to classify the prenatal ultrasound image as a CHD or NCHD. The curve stays in the upper left part of the graph, thus showing high sensitivity and specificity for various classification thresholds. The AUC value of 0.96 indicates very good diagnostic performance and discrimination between normal and abnormal fetal heart diseases. In conclusion, this overall figure validates the proposed AI-based system for prenatal detection and classification of CHD.

3.5.Risk Stratification Results

The ECHOFOCUS-CHD system was also able to provide automated risk stratification for fetal cardiac abnormalities. The risk levels for the various classes are listed below.

Table 4: Automated Risk Stratification

Risk Category	Number of Cases
Low Risk	108
Moderate Risk	42
High Risk	30
Critical Risk	20

As presented in Table 4, the automated risk stratification of fetal cardiac abnormality by the ECHOFOCUS-CHD system worked well according to the disease severity. Most (108) were considered to be low risk, meaning there were no or less severe cardiac issues. The model was able to differentiate the severity of congenital heart disease as a moderate number of cases were identified as moderate risk (42) and high risk (30). Moreover, 20 cases were classified as critical risk, showcasing the system's ability to detect serious fetal cardiac abnormalities that require immediate medical intervention. The results overall indicate the potential of the proposed framework to assist clinicians in prenatal risk assessment and early clinical decision making by means of an AI system.

4. DISCUSSION

In this current study, an AI-based autonomous framework to detect and stratify Critical Congenital Heart Disease (CHD) from prenatal ultrasound images, namely ECHOFOCUS-CHD, was developed and evaluated. The results showed that the model proposed, the Convolutional Neural Network (CNN), has high diagnostic accuracy, high sensitivity, high specificity and high classification rate in each group of CHD severity. The accuracy of 94.5% and the AUC value of 0.96 suggest that the proposed framework has the potential to successfully detect fetal cardiac abnormalities in prenatal ultrasound images.

4.1.Interpretation of Results

The findings of the study suggest that the proposed framework using AI successfully achieved the differentiation of normal fetuses from CHD cases and high reliability was achieved. High sensitivity value indicates that the system has a good ability to detect true positive CHD cases and high specificity indicates that the system has a good ability in identifying normal fetal cardiac condition. The matrix of errors also showed that the model is able to make a correct classification in most of the cases, with a few false-negative classifications of critical CHD cases. Clinically significant as early diagnosis of severe congenital heart defects can make a significant difference in the neonatal survival and therapy.

The risk stratification element of the framework also showed effective risk stratification of fetal cardiac abnormalities into groups of low, moderate, high and critical risk. This could be useful to clinicians for prenatal risk assessment, treatment planning and decision making during prenatal management.

4.2.Comparison with Existing Studies

Results of the present study are in line with the previous studies that have successfully shown the effectiveness of AI and deep learning in fetal cardiac imaging and fetal CHD detection.

Table 5: Comparison with Existing Studies

Author(s) & Year	Study Focus	Key Findings	Comparison with Present Study
Salih et al. (2023)[11]	Explainable AI in cardiac imaging	Highlighted the importance of interpretable AI models in cardiac diagnostics	The present study also supports AI-assisted cardiac diagnosis and demonstrates reliable classification performance
Sapitri et al. (2023)[12]	Real-time deep learning detection in fetal ultrasound videos	Achieved effective real-time fetal cardiac object detection using deep learning	Similar to their findings, the proposed model showed high detection accuracy for CHD cases
Tan et al. (2020)[13]	Automated detection of congenital heart disease in fetal ultrasound screening	Reported successful automated CHD detection using AI techniques	The current study extends this work by including automated risk stratification along with disease detection
Wong et al. (2021)[14]	Deep learning-based fetal echocardiography segmentation	Demonstrated improved segmentation and multiclass classification performance	The present study similarly applied CNN-based classification for multiple CHD categories
Xin et al. (2026)[15]	Review of prenatal ultrasound diagnosis of fetal CHD	Emphasized the growing role of AI in improving prenatal CHD diagnosis	The findings of the present study further support the integration of AI in prenatal cardiac screening

This study adds to the body of knowledge by integrating automated disease detection with risk stratification into a unified AI framework. ECHOFOCUS-CHD offers a more comprehensive prenatal screening approach than previous studies, leveraging the ability to classify the disease severity and generate risk categories for clinical support.

4.3.Implications of Findings

The findings of this study have important clinical and technological implications. The proposed ECHOFOCUS-CHD framework can help healthcare providers to better detect CHD in the first trimester. By leveraging AI in prenatal ultrasound screening, the technology has the potential to minimize diagnostic variations, enhance screening effectiveness, and aid clinicians in low-resource healthcare environments where fetal cardiology expertise is less readily available.

The automated risk stratification system could also help to identify high-risk pregnancies for further medical intervention and early treatment. Moreover, deep learning models could play a role in developing precision medicine and AI-driven healthcare systems in prenatal diagnostics.

4.4.Limitations of the Study

The study has several limitations, although the results are promising. First, the number of prenatal ultrasound images in the dataset was relatively small, with 200 images, which could restrict the applicability of the results. Second, the study was retrospective and the variance in image quality could have had an impact on the performance of the models. Third, there was a limited number of CHD categories used in evaluating the model, and it might not be reflective of the total range of congenital cardiac abnormalities. Moreover, external clinical validations at various healthcare centers were not carried out.

4.5.Suggestions for Future Research

Further research is needed to increase the amount of data and incorporate multi-center clinical data to enhance generalizability and robustness of the model. The integration of AI models with ultrasound imaging systems in real time could further improve the potential use of the technology in the clinic. Moreover, future research can investigate more sophisticated deep learning models like hybrid CNN-transformer architectures and explainable AI methods for enhancing interpretability and clinician trust.

Moreover, future studies could involve a larger and more diverse patient population, more categories of CHD, and prospective clinical trials to assess the real-world applicability of AI-driven prenatal cardiac screening systems in healthcare settings.

5. CONCLUSION

5.1. Summary of Key Findings

In the present study, an AI-based fully autonomous framework for detecting and stratifying Critical Congenital Heart Disease (CHD) from the prenatal ultrasound images, namely ECHOFOCUS-CHD, was successfully developed and evaluated. The proposed Convolutional Neural Network (CNN) model showed excellent diagnostic performance, with an overall accuracy of 94.5%, sensitivity of 92.8%, specificity of 95.6%, and an AUC of 0.96. The model successfully categorized fetuses into normal, mild, moderate and critical CHD categories. The accuracy and effectiveness of the proposed framework were also confirmed by the confusion matrix and ROC curve analyses. The system was also able to automatically classify the fetus into risk groups for cardiac anomalies, facilitating early prenatal diagnosis and clinical decision making.

5.2.Significance of the Study

The study highlights the growing importance of artificial intelligence and deep learning technologies in prenatal healthcare and fetal cardiology. The ECHOFOCUS-CHD framework is a proof of concept that shows the potential of AI-based prenatal ultrasound analysis to enhance the accuracy, consistency, and efficiency in the identification of CHD. Automated classification and risk stratification can help the clinician with early diagnosis, treatment planning and management of high-risk pregnancies. Additionally, the framework could prove to be useful in low-resource health care environments where access to experienced fetal cardiologists is scarce.

5.3.Final Thoughts and Recommendations

Based on the results of this study, AI-based prenatal diagnostic systems can contribute greatly to the improvement of fetal cardiac screening and diagnostic variability. The proposed framework showed promising results but larger and more diverse data sets are recommended to enhance the model's generalizability and its applicability in clinical settings. Real-time ultrasound integration, multi-center validation, and explainable AI techniques for improved clinical trust and adoption are also areas of future research that should be explored. In total, ECHOFOCUS-CHD is a significant contribution in the area of intelligent and reliable prenatal healthcare system for early detection of congenital heart disease.

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