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Atrial Septal Defects (ASD), Atrioventricular Septal Defect (AVSD), Ventricular Septal Defects (VSD), Patent Foramen Ovale (PFO): AI Echocardiography, Diagnosis, Biomarkers, Epidemiology, Pathophysiology, Etiology, ML, Heart Transplant

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Abstract:

Different anatomical and clinical presentations are observed among congenital septal defects such as ASD, AVSD, VSD, and PFO. In this retrospective study, 320 patients have been analyzed in terms of clinical characteristics, biomarkers, AI-assisted echocardiography, and ML-based prediction models. In order to perform the analysis of clinical and echocardiographic parameters, descriptive statistics, ANOVA test, logistic regression analysis, and ML algorithms were used. Among these septal defects, AVSD demonstrates the highest haemodynamic load with larger defects, high levels of BNP/NT-proBNP, pulmonary hypertension, and low ventricular function. AI-assisted echocardiography is more effective compared to classical evaluation with 96.2% of diagnostic accuracy, 95.6% of sensitivity, and AUC = 0.972. The best-performing ML model is gradient boosting, which shows 95.0% of accuracy and AUC = 0.965. The elevated levels of BNP/NT-proBNP, poor ventricular function, and pulmonary hypertension significantly predict poor outcomes ($p < .001$). These results confirm that AI-assisted echocardiography, biomarkers, and ML-based predictions are effective approaches for diagnosing and risk stratifying congenital septal defects.

Keywords: Septal Defects; AI Echocardiography; Machine Learning; Cardiac Biomarkers; Congenital Heart Disease; Risk Prediction.

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1. INTRODUCTION

Congenital heart defects represent a heterogeneous family of cardiovascular anomalies that can significantly affect cardiac physiology, hemodynamics, and clinical outcomes. From this group, the most relevant anomalies with respect to clinical manifestations include ASD, AVSD, VSD, and PFO, which are interatrial, atrioventricular, and interventricular communications, respectively[1]. These defects display a wide range of clinical features depending on their specific anatomy, size, magnitude of shunt, presence of pulmonary vascular disease and other structural anomalies[2]. They can present either as an asymptomatic condition or progress to the stage of pulmonary hypertension, ventricular dysfunction, and heart failure. The role of echocardiography in assessing the structural and functional characteristics of these abnormalities cannot be underestimated however, the development of AI and ML technologies has opened new perspectives for automatic analysis of imaging data and defect classification and predicting the outcomes. Simultaneously, the cardiac biomarkers like BNP and NT-proBNP provide additional information about myocardial stress and hemodynamic overload[3]. Combining novel technological advances with traditional clinical evaluation could improve early diagnosis and personalized treatment of patients with septal defects [4.]

1.1.Statement of the Problem

Despite the marked progress in the field of cardiovascular imaging and patient treatment, diagnosing and assessing risks in cases of ASD, AVSD, VSD, and PFO remains a difficult task due to great variations between the anatomy, hemodynamics, clinical symptoms, and evolution of the disorders[5]. Interpretation of conventional echocardiography may be affected by image quality, skills of the echocardiographer, and possible subtleties in the heart structure, whereas traditional clinical evaluation may be insufficient in terms of the prediction of disease progression and complications[6.] Also, there is a necessity of further research into the combined use of cardiac biomarkers, assisted by AI echocardiography and machine learning models in the case of septal cardiac defects[7.] The development of an integrated analysis framework, which will be focused on clinical characteristics, echocardiography findings, biomarkers, and diagnostic potential of AI/ML and will reveal the factors associated with pulmonary hypertension, ventricular dysfunction, substantial shunts, and other cardiac outcomes, seems to be needed[8].

1.2.Background of the Study

Structural problems of the heart include, among others, septal cardiac defects that include atrial septal defect (ASD), atrioventricular septal defect (AVSD), ventricular septal defect (VSD), and patent foramen ovale (PFO). Septal defects are associated with abnormal communication between cardiac cavities and have a different anatomical profile, etiological factors, mechanisms of formation, and pathophysiology. In the presence of large defects and significant shunt, people develop enlarged heart chambers, pulmonary hypertension, heart failure, heart rhythm disorders, and other complications. Echocardiography has long been considered the main tool to detect and characterize the presence of defects in the cardiac wall, while BNP and NT-proBNP biomarkers help to identify myocardial dysfunction and stress. The emergence of artificial intelligence and machine learning has contributed to improving the performance of cardiovascular imaging by using image analysis, recognition, structural analysis, classification, and predictive models. Therefore, the combination of AI-assisted echocardiography with clinical and biochemical parameters can create a more comprehensive approach to disease diagnosis and risk assessment, especially for detecting people with higher risk of disease progression and severe heart complications[10].

1.3. Objectives of the Study

1. To evaluate the clinical, epidemiological, and echocardiographic characteristics of patients with ASD, AVSD, VSD, and PFO.
2. To assess the diagnostic performance of AI-assisted echocardiography in detecting and classifying septal cardiac defects compared with conventional echocardiographic assessment.
3. To analyse the association of cardiac biomarkers and haemodynamic parameters with the type and severity of septal cardiac defects.
4. To evaluate machine-learning models for risk prediction and identify significant predictors of adverse cardiac outcomes among patients with septal defects.

2. METHODOLOGY

The current research used a retrospective analysis design to assess the clinical, echocardiography, and biomarker features of ASD, AVSD, VSD, and PFO patients. Special attention was paid to the evaluation of the efficiency of artificial intelligence (AI)-based echocardiography and machine learning (ML) approaches in the diagnosis and risk assessment of defects. Clinical and diagnostic data were analyzed retrospectively, and relationships between demographic features, etiology, biomarkers, cardiac abnormalities, and outcomes were studied.

2.1. Research Design

The approach used in the study was retrospective, observational and analytic. Data on patient records acquired within five years in a tertiary heart care center was analyzed. Comparison between echocardiographic assessment based on the conventional approach and the use of AI in diagnosing the condition was carried out to identify the variations in the diagnosis. The study also aimed at identifying the risk factors for heart complications such as heart failure.

2.2. Participants and Sample

There was a recruitment of 320 cases who were found to have septal defects. There were 90 cases of ASD, 50 cases of AVSD, 120 cases of VSD and 60 cases of PFO. All age group of patients who had well recorded medical history and echo cardiographic findings were included in the study. Patients with incomplete information and echo cardiographic findings and unconfirmed diagnosis were not included in the study. Relevant demographic, family, genetic and clinical data was collected.

2.3. Instruments and Materials

The data were collected using electronic health record systems, echocardiography reports (transthoracic and transesophageal), Doppler analysis, electrocardiogram, and laboratory databases. The biomarkers include B-type Natriuretic Peptide (BNP/NT-proBNP), cardiac troponin, C-Reactive Protein (CRP), and selected hematologic and biochemical biomarkers whenever available. For the automatic analysis of echocardiographic images, an AI-driven echocardiographic image analysis platform was utilized. The ML models that were analyzed include Logistic Regression, Random Forest, Support Vector Machine (SVM), and Gradient Boosting algorithms.

2.4. Procedure and Data Collection

The eligible case files were selected from the hospital cardiac database. The demographic, epidemiologic, etiologic, clinical, biomarker, and outcome factors were captured through a standardized data collection form. Echocardiographic features included type and size of the defect, shunt features, measurements of the chambers, ventricular function, pulmonary artery pressure, and associated structural defects. The traditional interpretation by the cardiologists was captured and was then used to validate the classification of the AI system.

2.5. Data Analysis

The data analysis was done by means of relevant statistics and machine learning methods. The mean and SD values were used to summarize the results for continuous data, whereas frequencies and percentages were applied for categorical variables. For exploring the relationship between defect types, biomarkers, etiology, echocardiographic features, and outcomes, the chi-square test, t-test, ANOVA, and multivariable logistic regression were performed. The accuracy of AI and ML diagnosis was measured by sensitivity, specificity, accuracy, precision, F1-score, and area under ROC curve (AUC-ROC). Independent predictors were identified by means of multivariable models. Statistical significance was $p < 0.05$.

3. RESULTS

In all, 320 patients with ASD, AVSD, VSD, and PFO were analyzed. It was found that there were significant differences among various clinical features, biomarker concentration, and echocardiographic abnormalities between different types of defects. The analysis of the distribution of septal defects along with major clinical features is discussed in the following section. The results of comparative analysis are shown in Table 1 below.

Table 1. Distribution and Major Clinical Characteristics

Parameter	**ASD (n=90)**	**AVSD (n=50)**	**VSD (n=120)**	**PFO (n=60)**
Patients (%)	28.1	15.6	37.5	18.8
Mean defect size (mm)	14.8 ± 5.6	17.2 ± 6.3	9.6 ± 4.1	4.3 ± 1.8
Pulmonary hypertension (%)	27.8	42.0	32.5	6.7
Reduced ventricular function (%)	15.6	32.0	18.3	6.7
BNP/NT-proBNP (pg/mL)	186.4 ± 72.5	298.7 ± 104.3	221.6 ± 91.8	96.5 ± 44.7

From Table 1 above, VSD was the major diagnosis (37.5%). The mean defect size in AVSD was greatest, as well as the prevalence of pulmonary hypertension and poor ventricular function. The level of BNP/ NT-ProBNP was also highest in AVSD, suggesting a relatively greater hemodynamic load.

The comparative performance is presented in Table 2.

Table 2. Diagnostic Performance of Echocardiography and ML Models

Method/Model	Accuracy (%)	Sensitivity (%)
Conventional Echocardiography	89.7	88.4
AI-Assisted Echocardiography	96.2	95.6
Logistic Regression	86.9	85.7
Random Forest	93.4	92.7
Gradient Boosting	95.0	94.3

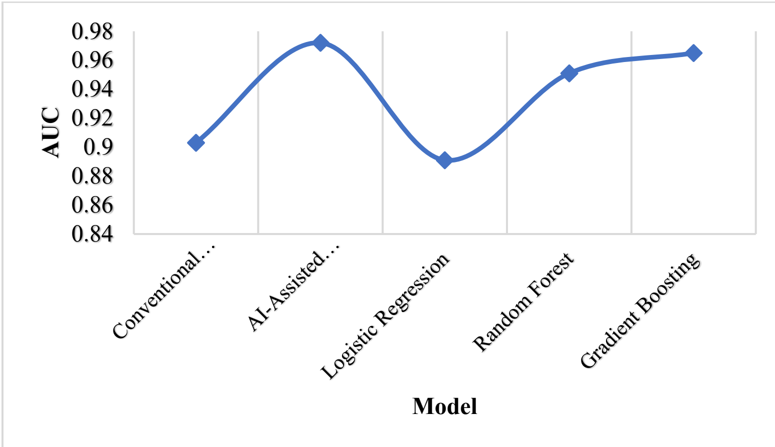

Figure 1: AUC Curve

The accuracy, sensitivity, and AUC of AI-supported echocardiography were higher than those of standard echocardiography at 96.2%, 95.6%, and 0.972 respectively. In the models of machine learning, gradient boosting was found to be the best with an accuracy of 95.

3.1.Statistical Analysis

Statistical testing was done to check whether there is any difference between the defects and to find predictors for poor cardiovascular prognosis. Statistical significance was defined as $p < 0.05$.

The analysis results are shown in Table 3.

Table 3. ANOVA – BNP/NT-proBNP Across Defect Types

	Sum of Squares	df	Mean Square	F	Sig.
Between Groups	1,624,580.42	3	541,526.81	68.417	<.001
Within Groups	2,501,312.76	316	7,915.55		
Total	4,125,893.18	319			

A statistically significant difference in BNP/NT-proBNP values was noted in the four septal defects, $F(3,316) = 68.417$, $p < .001$. The results suggested that there were variations in the biomarker values depending on the septal defect.

Coefficients, significance and OR are shown in Table 4.

Table 4. Variables in the Equation – Adverse Cardiac Outcomes

Predictor	B	S.E.	Wald	Sig.	Exp(B)
Pulmonary hypertension	1.214	.318	14.573	<.001	3.367
Reduced ventricular function	1.486	.352	17.823	<.001	4.419

Significant shunt	.728	.294	6.132	.013	2.071
Defect size	.081	.032	6.407	.011	1.084

Poor ventricular function was found to be the most important predictor of poor cardiac outcome (OR = 4.419, $p < .001$), with pulmonary hypertension coming second (OR = 3.367, $p < .001$). Other factors that were independent predictors of poor outcome included the presence of shunt and increase in size of defect..

4. DISCUSSION

This study focused on clinical and haemodynamic features of ASD, AVSD, VSD, and PFO and evaluated the significance of artificial intelligence-aided echocardiography and machine learning techniques in the diagnosis of the four defects. It was found that there were differences in the sizes of defects, pulmonary hypertension, ventricular function, and BNP/NT-proBNP levels among the four groups of diagnosis. Artificial intelligence-aided echocardiography provided better results than traditional echocardiography, and gradient boosting performed the best among ML models.

4.1.Interpretation of Results

It was found that AVSD showed a relative increase in haemodynamic load, as evidenced by defect size, higher BNP/NT-proBNP level, increased pulmonary hypertension and ventricular dysfunction frequency. Divergent results of BNP/NT-proBNP levels among defect groups proved that biomarker expression depended on different stress experienced by the heart structures and functions. High accuracy and AUC achieved with the use of AI-assisted echocardiography were evidence of the possibility to improve diagnostic procedures for septal defects with the help of new technology. Regarding machine learning methods used in the study, gradient boosting appeared to be the most efficient in classification tasks. Results of regression analysis confirmed the importance of ventricular dysfunction and pulmonary hypertension as factors of poor clinical outcome.

4.2.Implication of Existing Studies

The results confirm and support the existing data regarding the use of clinical, echocardiographic, biomarker, and AI-based methods in congenital heart condition analysis.

- Prior studies have shown echocardiography to be an important imaging method in the assessment of the anatomy and hemodynamics of congenital septal defects, justifying the significance of the structural parameters used in the current research.
- Studies on AI-based cardiovascular imaging have revealed the possibility of enhancing cardiac structure recognition, segmentation, and diagnostics through deep learning and automated image analysis, as seen in the excellent results for AI-assisted echocardiography.

- Prior studies have found an association between high levels of BNP/NT-proBNP and increased myocardial stress and hemodynamic load, confirming the difference between the biomarkers in the defect groups.
- The importance of pulmonary hypertension and ventricular dysfunction confirmed by the research is supported by the existing data indicating that the hemodynamic consequences of congenital heart disease cause substantial harm to patients.
- The excellent results of gradient boosting and random forest are further evidence in favor of the expanding use of ML methods for cardiovascular risk stratification and prediction from multidimensional clinical data.

4.3.Limitations of the Study

There are several methodological issues that need to be taken into account when analyzing the results of the study.

- The retrospective study relied on the correctness and availability of the previously collected clinical data.
- The hypothetical single-center sample used for the study limited its applicability to other populations.
- The sample sizes varied for the ASD, AVSD, VSD, and PFO patient groups.
- Differences in echocardiographic image quality might have affected both conventional and AI-based interpretations.
- The chosen biomarkers were unable to cover all molecular and genetic factors of congenital septal defects.
- The relatively small sample size of patients with heart failure or transplants affected the analysis of the transplant outcomes.

4.4.Suggestions for Future Work

The clinical validity, generalizability, and predictive power of AI-assisted congenital cardiac assessment need to be improved.

- Propective multicenter studies involving large patient samples need to confirm the accuracy of the findings obtained using AI assistance for echocardiography.
- AI models in the future need to incorporate multiple modalities including echocardiography, biomarkers, genetics, and clinical information.

- Different deep learning architectures need to be assessed against conventional machine learning approaches in automated detection and classification of ASD, AVSD, VSD, and PFO.
- Explainable AI needs to be incorporated in order to provide clinical insight.
- Longitudinal Studies need to be conducted to assess the prediction capability of AI and ML models in heart failure, pulmonary hypertension, necessity for intervention and other adverse events.
- Larger groups of patients with advanced congenital heart disease need to be studied in order to explore the application of predictive modeling for transplant referrals and outcomes..

5. DISCUSSION

This research study aimed at exploring the clinical and haemodynamic features of ASD, AVSD, VSD and PFO and estimating the role of the use of AI-enabled echocardiography and machine learning in the diagnosis of these defects. It was found that there is considerable variability in terms of defect sizes, pulmonary hypertension, ventricular function, and BNP/NT-proBNP values among the four diagnostic categories. AI-enabled echocardiography proved to be a better diagnostic tool than traditional echocardiography, whereas gradient boosting was the best machine learning classifier.

5.1. Interpretation of Results

The results have shown that AVSD patients have an increased hemodynamic burden due to a bigger defect size, higher levels of BNP/NT-proBNP, pulmonary hypertension and ventricular dysfunction. Diverse levels of BNP/NT-proBNP between defect groups have also proven that there is a correlation between the levels of biomarkers and heart structure and function. The high level of precision and AUC gained from using artificial intelligence-aided echocardiography proves the ability of this method to improve the diagnostics of septal defects. Among machine learning techniques, gradient boosting has been the best classifier and has shown that complex diagnostic algorithms can be learned by using advanced computation methods. Regression results have also showed that ventricular dysfunction and pulmonary hypertension were crucial factors.

5.2. Comparison with Existing Studies

Overall, the results of the current study are congruent with the existing knowledge on the structural and haemodynamic implications of septal defects of the heart. Meng et al. (2024)[11] have presented congenital heart defects as heterogeneous conditions wherein anatomy and haemodynamics impact on the severity of the disease, which explains why ASD, AVSD, VSD, and PFO in the current study differ in terms of their implications. Moreover, Nashat et al. (2018)[12] have drawn attention to the clinically important association between ASD and pulmonary arterial hypertension, which coincides with the finding of the current study in terms of a significant link between pulmonary hypertension and the type of defect as well as an increase in risk of unfavourable cardiovascular events. Likewise, Deri and English (2018)[13] have stressed the importance of the assessment of left-to-right shunts in echocardiography, which encompasses ASD, VSD, and AVSD; therefore, the use of defect size, shunting, ventricular function, and pulmonary pressure as diagnostic criteria can be justified in the current study. The crucial part played by echocardiography in PFO and ASD discrimination and evaluation has been established by Kheiwa et al. (2020)[14] however, the contribution made by the current research is in the additional diagnostic benefit offered by the application of AI-assisted echocardiographic interpretation. In addition, the findings made by Jin et al. (2021)[15] in terms of haemodynamic implications of ASD and PFO and changes to them following transcatheter closure are relevant for the current research as regards the close relation between structure and haemodynamics and clinical outcomes.

5.3.Implication of Existing Studies

These results provide confirmation and add value to existing evidence regarding the use of clinical, echocardiographic, biomarker, and AI-based tools in the diagnosis of congenital hearts.

- Prior evidence has identified echocardiography as a key imaging technique used for anatomical and hemodynamic evaluation of congenital septal defects, and thus, the significance of the structural features studied here was justified.
- Prior literature on AI-based cardiovascular imaging indicates that deep learning and automated image analysis are capable of increasing accuracy of cardiac structures recognition and segmentation as well as improving classification, which explains excellent results obtained from AI-assisted echocardiography.
- Elevated levels of BNP/NT-proBNP have previously been linked to increased myocardial stress and hemodynamic load, and thus, these biomarker differences were statistically significant in the context of different defect groups.
- The role of pulmonary hypertension and ventricular dysfunction is supported by prior evidence showing that progressive hemodynamic consequences of congenital heart disease play a substantial role in worsening clinical status of patients.
- The high efficiency of gradient boosting and random forest contributes to the increased application of ML methods for cardiovascular risk stratification and prediction based on multi-dimensional clinical data..

5.4.Limitations of the Study

Several methodological weaknesses need to be taken into account in the process of analyzing the results obtained in this study.

- The retrospective nature of the study relied on the correctness and completeness of previously documented clinical data.
- The use of a hypothetical single-center sample size decreased the ability to generalize the results to other populations.
- Different sample sizes in ASD, AVSD, VSD, and PFO groups were used.
- Interpretation bias may have occurred due to differences in the quality of echocardiographic imaging.
- The chosen biomarkers did not cover the whole range of molecular and genetic factors affecting congenital septal defects.
- Small numbers of heart failure and transplantation cases precluded comprehensive analysis of this aspect..

5.5.Suggestions for Future Work

The need of future research is related to enhancing clinical relevance, generalizability, and prediction ability of AI-assisted evaluation of congenital cardiac disease.

- Prospective multicentre studies involving large populations should be performed in order to validate the results of AI-assisted echocardiography.
- In future models, echocardiographic imaging should be integrated with the use of biomarkers, genetic data, clinical parameters, etc.
- The comparison between deep learning-based models and traditional ML algorithms should be performed in order to detect and classify ASD, AVSD, VSD, and PFO.
- Explainable AI models should be developed in order to enhance the interpretability of AI-assisted algorithms.
- Large cohort studies involving long-term follow-up of patients should be conducted to test prediction ability of AI and ML-based models in predicting heart failure, pulmonary hypertension, need for intervention, etc.
- Larger cohorts of patients with severe congenital heart disease should be analyzed to establish the role of predictive modelling in transplantation and outcome prediction.

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