

CLINICAL CHARACTERISTICS, MANAGEMENT STRATEGIES, AND CLINICAL
OUTCOMES OF ADULTS WITH CONGENITAL HEART DISEASE AT A
TERTIARY CARE HOSPITAL IN PAKISTAN

Dr. Jawad Ahmad

4th Year PG Internal Medicine. jawadahmad1587@gmail.com

Dr. Rani Aqsa Islam

MBBS from QAMC Bahawalpur. dr.rani14@gmail.com

Sartaj Ali

Lecture in Surgical technology at KMU IHS Kohat. sartajaliwazir2002@gmail.com

Zia Ur Rahman

Cardiology Technologist at Rehman medical institute Peshawar. Xxiaurrahman@gmail.com

Ghulam Murtaza

Cardiology Technologist. ghulammurtaza1858@gmail.com

Shariq Amin

Cardiology Technologist. shariqamin2020@gmail.com

Aitazaz Rafi

MPH Scholar Khyber Medical University, Peshawar, Healthcare Quality Professional (PIQC)
Shoukat Khanum Cancer Hospital and Research Centre Peshawar. aitazazrafi@gmail.com

Shoukat Ali*

Lecturer Cardiology at Iqra National University. Corresponding Author Email:
shaukatali9343193@gmail.com

Abstract

Author Details

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Corresponding E-mails & Authors*:

Shoukat Ali*

shaukatali9343193@gmail.com

Objective: To assess the clinical presentation, management approaches, and short-term outcomes among adult patients with congenital heart disease (ACHD) presenting to a tertiary care hospital in Peshawar, Pakistan.
Methodology: This cross-sectional study was conducted in the Department of Pediatric Cardiology and Intensive Care Unit (ICU) at Lady Reading Hospital, Peshawar, Pakistan, from March 2025 to November 2025. Patients of either gender, aged 18 years or older, with a confirmed diagnosis

of congenital heart disease were included in the study. Data regarding demographic characteristics, clinical presentation, type of congenital heart defect, therapeutic interventions, and clinical outcomes were collected and analyzed. Outcomes were

assessed in terms of intra-operative or postoperative mortality and postoperative complications. **Results:** A total of 129 patients with adult congenital heart disease were included, of whom 68 (52.7%) were male. The median age of the patients was 22.00 years. Ten (7.8%) patients were newly diagnosed with congenital heart disease during the study period. The most frequently observed congenital heart defects were tetralogy of Fallot (TOF), atrial septal defect (ASD), and ventricular septal defect (VSD), accounting for 32 (24.8%), 25 (19.4%), and 22 (17.1%) cases, respectively. The most common presenting complaints were shortness of breath, cyanosis, and palpitations, reported in 120 (93.0%), 48 (37.2%), and 44 (34.1%) patients, respectively. Cardiac catheterization was the most frequently performed intervention, carried out in 41 (31.8%) patients. Total correction, VSD closure, and ASD closure were performed in 9 (7.0%), 7 (5.4%), and 7 (5.4%) patients, respectively. No intra-operative or postoperative mortality was recorded. The most frequently documented postoperative complications were left ventricular dysfunction in 39 (30.2%), right ventricular dysfunction in 23 (17.9%), biventricular dysfunction in 6 (4.7%), and ventricular hypertrophy in 3 (2.3%) patients. **Conclusion:** Tetralogy of Fallot, atrial septal defect, and ventricular septal defect were the most commonly encountered congenital heart diseases among adults in this study. A considerable proportion of patients required surgical or catheter-based interventions. Despite the complexity of the underlying cardiac conditions, favorable short-term outcomes were observed, with no intra-operative or postoperative mortality reported. **KEYWORDS:** Adult congenital heart disease, Atrial septal defect, Cardiac catheterization, Congenital heart disease, Tetralogy of Fallot, Ventricular septal defect.

INTRODUCTION

Over the past several decades, substantial changes have occurred in the epidemiology, diagnosis, and management of congenital heart disease (CHD). Historically, the prevalence of CHD was estimated to be approximately 1 in every 100 live births.¹ By 2000, reported estimates indicated a prevalence of 11.89 per 1,000 children, 4.09 per 1,000 adults, and 5.78 per 1,000 individuals in the general population. Subsequent estimates in 2010 demonstrated a prevalence of approximately 6.1 per 1,000 among adults.² A considerable proportion of adults with congenital heart disease (ACHD) have

moderate- to high-complexity cardiac defects, highlighting the ongoing clinical burden associated with these conditions.^{3,4}

Advances in pediatric cardiac surgery, interventional cardiology, and hybrid treatment approaches have markedly improved the survival of patients with CHD. Improved diagnostic techniques and advances in surgical and postoperative care have allowed a growing number of children with congenital cardiac abnormalities to survive into adolescence and adulthood. Survival among individuals with many forms of CHD has increased substantially, with reported survival rates approaching 97% for several congenital cardiac conditions.⁵ Approximately 60% of CHD cases are diagnosed and managed during the first year of life, while a further proportion is identified during childhood and adolescence, up to approximately 16 years of age.⁶ Nevertheless, around 10% of individuals may remain undiagnosed until adulthood.⁶

The increasing number of adults living with CHD has created an expanding need for long-term specialized cardiac care. Many patients who undergo corrective procedures during infancy or childhood require additional interventions later in life because of residual defects, progressive cardiac dysfunction, or postoperative complications.⁷ Consequently, improvements in childhood survival have contributed substantially to the growing population of adults living with congenital cardiac disease. This changing clinical landscape emphasizes the importance of continuous, multidisciplinary cardiac care throughout the patient's lifespan.

The clinical consequences of CHD during adolescence and adulthood are particularly important. Delayed diagnosis or correction of significant congenital cardiac lesions may result in progressive and potentially irreversible cardiovascular changes. In some patients, late presentation can lead to pulmonary vascular disease, ventricular dysfunction, arrhythmias, cyanosis, or other complications that may limit the feasibility of definitive corrective treatment.⁸ Therefore, early recognition, appropriate intervention, and continued follow-up are essential for reducing disease-related morbidity and improving long-term outcomes among patients with ACHD.

In Pakistan, limited evidence is available regarding the clinical characteristics, patterns of presentation, management strategies, and outcomes of adults living with congenital heart disease. Data from tertiary care hospitals in Khyber Pakhtunkhwa, particularly

regarding the spectrum of ACHD and the outcomes of surgical and catheter-based interventions, remain relatively scarce. Understanding the clinical profile and treatment outcomes of these patients is important for identifying areas requiring improved diagnostic services, timely referral, specialized intervention, and long-term follow-up.

Therefore, the present study was conducted at Lady Reading Hospital, Peshawar, to evaluate the clinical presentation, types of congenital cardiac defects, management approaches, and short-term outcomes among adult patients with congenital heart disease. The findings may contribute to a better understanding of the local burden of ACHD and may assist healthcare professionals in improving the delivery and quality of cardiac care for this growing patient population.

MATERIAL AND METHOD

Study Design and Setting

This cross-sectional study was conducted in the Department of Pediatric Cardiology and Intensive Care Unit (ICU) at Lady Reading Hospital, Peshawar, Pakistan, from March 2025 to November 2025.

Ethical Considerations

Ethical approval was obtained from the Institutional Ethical Review Board of Lady Reading Hospital, Peshawar, before commencement of the study. Written informed consent was obtained from all participants after explaining the purpose and procedures of the study. Confidentiality of participants' information was maintained throughout the research process.

Inclusion Criteria

Patients of either gender, aged 18 years or older, with a confirmed diagnosis of congenital heart disease (CHD) were included. Both previously diagnosed and newly diagnosed cases of CHD were considered, provided that the diagnosis was confirmed through transthoracic echocardiography.

Exclusion Criteria

Patients with acquired heart disease, cardiomyopathy, or congenital arrhythmias were excluded from the study.

Diagnosis of Congenital Heart Disease

The diagnosis of CHD was established using two-dimensional and color Doppler transthoracic echocardiography. The echocardiographic assessment included M-mode, two-dimensional imaging, color Doppler, pulsed-wave Doppler, and continuous-wave Doppler techniques. Congenital heart disease was defined as a gross structural abnormality of the heart or intrathoracic great vessels that is actually or potentially of functional significance, excluding abnormalities involving the systemic arteries and veins.

Data Collection

Data were collected using a structured data collection proforma. Information regarding demographic characteristics, including age, gender, and body mass index (BMI), was recorded. Clinical information included electrocardiographic (ECG) findings, age at diagnosis of CHD, maternal history of congenital heart disease, presenting signs and symptoms, and CHD-related complications. Echocardiographic findings, New York Heart Association (NYHA) functional classification, and details of medical, surgical, and catheter-based interventions were also documented.

BMI was categorized according to predefined criteria, with a BMI below 23 kg/m² considered within the normal range. All patients received treatment according to the established institutional protocols. Details regarding the type of surgical or catheter-based intervention performed during the study period were recorded.

Outcome Measures

The primary outcomes were intra-operative mortality, postoperative mortality, and postoperative complications. Complications occurring following surgical or interventional procedures were documented and categorized according to the clinical findings observed during the hospital stay.

Statistical Analysis

Data were entered, processed, and analyzed using IBM SPSS Statistics, version 26.0. Categorical variables, including gender, residential characteristics, marital status, echocardiographic findings, clinical signs and symptoms, maternal history of CHD, CHD-related complications, NYHA functional class, interventions, and clinical outcomes, were presented as frequencies and percentages. Quantitative variables, including age and BMI, were expressed as median and interquartile range (IQR). The Chi-square test or

Fisher's exact test was used for comparison of categorical variables, as appropriate. A p-value of <0.05 was considered statistically significant.

Results:

A total of 129 patients with adult congenital heart disease (ACHD) were included in the study. Among these, 68 (52.7%) were male. The median age of the participants was 22.00 years (IQR: 20.00–28.50 years), while the median age at the time of CHD diagnosis was 5.00 years (IQR: 0.50–19.50 years). The median BMI was 17.57 kg/m² (IQR: 14.95–20.24 kg/m²).

The most frequently identified congenital heart defects were tetralogy of Fallot (TOF), atrial septal defect (ASD), ventricular septal defect (VSD), and patent ductus arteriosus (PDA), accounting for 32 (24.8%), 25 (19.4%), 22 (17.1%), and 10 (7.8%) patients, respectively. Right axis deviation was the most frequently observed electrocardiographic abnormality, present in 54 (41.9%) patients. The baseline demographic and clinical characteristics of the ACHD patients are presented in **Table I**.

Regarding clinical presentation, shortness of breath was the most common presenting complaint, reported by 120 (93.0%) patients, followed by cyanosis in 48 (37.2%), palpitations in 44 (34.1%), and chest pain in 39 (30.2%) patients.

A significant gender-based difference was observed among patients with different types of CHD. ASD was significantly more frequent among females, with females accounting for 72.0% of ASD cases compared with 28.0% among males ($p=0.006$). In contrast, VSD was significantly more common among males, who constituted 72.7% of cases compared with 27.3% among females ($p=0.039$). All patients diagnosed with coarctation of the aorta (CoA) and pulmonary stenosis were male, demonstrating a statistically significant association with gender ($p=0.031$).

Patients with TOF were significantly younger compared with patients having other types of CHD ($p=0.041$). Conversely, ASD was significantly more prevalent among patients belonging to older age groups ($p<0.001$). The associations between different CHD types, gender, age, and newly diagnosed cases.

Overall, 92 (71.3%) patients underwent surgical or interventional procedures during the study period. Cardiac catheterization was the most frequently performed intervention, accounting for 41 (31.8%) cases. Total correction was performed in 9 (7.0%) patients,

while VSD closure and ASD closure were performed in 7 (5.4%) patients each. PDA device closure was performed in 5 (3.9%) patients. **No intra-operative or postoperative mortality was recorded.** The distribution of the various surgical and interventional procedures performed among ACHD patients is illustrated in **Figure 2**.

Postoperative complications were also assessed. Left ventricular dysfunction was the most frequently observed complication, occurring in 39 (30.2%) patients, followed by right ventricular dysfunction in 23 (17.9%), biventricular dysfunction in 6 (4.7%), and ventricular hypertrophy in 3 (2.3%) patients.

Conclusion and Discussion

The evaluation and selection of patients with unrepaired adult congenital heart disease (ACHD) for corrective or palliative intervention remains challenging because of the considerable variation in disease severity, anatomical complexity, and clinical presentation. In the present study, the median age of the patients at presentation was 22.00 years (IQR: 20.00–28.50 years), whereas the median age at diagnosis was 5.00 years (IQR: 0.50–19.50 years). These findings are relatively comparable with previously published international data. A study from Mexico reported a median age of 36 years (IQR: 28–47 years) among patients with ACHD, while a study from Iran reported a mean age of 31.05 years.^{10,11} Similarly, data from Italy demonstrated a median age of 25.5 years, which was relatively close to the findings of the present study.¹²

Delayed diagnosis of congenital heart disease remains an important concern, particularly in developing countries, where socioeconomic limitations, inadequate access to healthcare, and limited availability of screening services may contribute to late recognition of congenital cardiac abnormalities.¹³ In the current study, 7.8% of patients were newly diagnosed with CHD during adulthood. Delayed diagnosis may result in the progression of cardiac disease and may increase the complexity of subsequent surgical or interventional management. Patients presenting late may have developed significant structural or functional cardiac abnormalities by the time they reach specialized cardiac centers.¹⁴ Furthermore, inadequate referral pathways and limited access to specialized congenital cardiac services may contribute to delays in definitive treatment.

The clinical presentation of ACHD varies according to the type and severity of the underlying cardiac defect. In our study, shortness of breath was the most common

presenting symptom, reported in 93.0% of patients, followed by cyanosis (37.2%), palpitations (34.1%), and chest pain (30.2%). Similar clinical manifestations have been reported in community-based studies, including research from Korea, where symptoms such as dyspnea, cyanosis, fatigue, and chest discomfort were associated with congenital heart disease.¹⁵ These symptoms may reflect the hemodynamic consequences of congenital cardiac lesions and demonstrate the diverse clinical burden experienced by adults living with CHD.

The distribution of congenital cardiac defects in the present study showed that tetralogy of Fallot (TOF), atrial septal defect (ASD), ventricular septal defect (VSD), and patent ductus arteriosus (PDA) were the most frequently encountered lesions, accounting for 24.8%, 19.4%, 17.1%, and 7.8% of cases, respectively. Although ASD is generally reported as one of the most prevalent forms of CHD among adults worldwide,¹⁶ local pediatric data have identified VSD and TOF among the most frequently observed congenital cardiac lesions.¹⁷ This difference may indicate variations in the spectrum of congenital heart disease between pediatric and adult populations and may also reflect differences in survival, referral patterns, diagnostic practices, and access to corrective treatment.

The pattern observed in our study also differs from findings reported from Canada, where coarctation of the aorta, partial anomalous pulmonary venous return (PAPVR), TOF, and Ebstein's anomaly were among the most frequently reported congenital cardiac lesions in adults.¹⁸ Such differences may reflect geographical, demographic, genetic, healthcare-access, and referral-related variations in the distribution of ACHD worldwide.

Several congenital heart defects demonstrate differences in their prevalence according to gender.¹⁹ Lesions involving the inflow portions of the heart, particularly ASD and Ebstein's anomaly, have generally been reported more frequently among females.^{20,21} Consistent with these observations, ASD was significantly more common among females in our study, with females accounting for 72.0% of ASD cases compared with 28.0% among males ($p=0.006$). Conversely, VSD demonstrated a significant male predominance, with males representing 72.7% of cases compared with 27.3% among females ($p=0.039$). In addition, all patients diagnosed with coarctation of the aorta and

pulmonary stenosis in our study were male, with a statistically significant association between these lesions and gender ($p=0.031$).

Previous studies have also suggested a male predominance for certain outflow tract abnormalities, including aortic stenosis and transposition of the great arteries.²² The observed gender differences may be related to differences in embryological development, genetic factors, and the developmental pathways involved in cardiac morphogenesis. Data from the CONCOR registry have suggested that the overall gender distribution among adults with congenital heart disease is broadly comparable with the gender distribution observed at birth.²³ Nevertheless, the apparent gender-specific distribution of individual congenital cardiac defects warrants further investigation using larger, multicenter studies involving local populations.

Interventional cardiology has become an increasingly important component of comprehensive ACHD management. Catheter-based procedures provide therapeutic options for selected patients and may reduce the need for conventional surgery in appropriately selected lesions. In the present study, a substantial proportion of patients required surgical or catheter-based intervention, reflecting the complexity of the disease burden among adults presenting to a tertiary cardiac care facility. Appropriate patient selection, advanced imaging, experienced multidisciplinary teams, and access to specialized congenital cardiac services remain important factors in achieving favorable outcomes.

Overall, the present study demonstrated that TOF, ASD, VSD, and PDA were the predominant congenital heart defects among adult patients. Shortness of breath, cyanosis, palpitations, and chest pain were the most frequently reported presenting symptoms. Significant gender-based differences were observed, with ASD being more common among females and VSD more frequently observed among males. A substantial proportion of patients required surgical or catheter-based interventions, while no intra-operative or postoperative mortality was documented. These findings highlight the importance of early diagnosis, timely referral, and specialized multidisciplinary management of adults with congenital heart disease.

CONCLUSION

The study demonstrated that **TOF, ASD, VSD, and PDA were the most frequently encountered congenital heart defects among adults**. Shortness of breath, cyanosis, palpitations, and chest pain represented the major presenting symptoms. ASD showed a significant female predominance, whereas VSD was significantly more frequent among males. A considerable proportion of patients required surgical or catheter-based interventions, with favorable short-term outcomes and no recorded intra-operative or postoperative mortality.

RECOMMENDATIONS

Further research should focus on the **long-term outcomes of adults with congenital heart disease**, including survival, recurrent interventions, cardiac function, complications, quality of life, and functional status. Multicenter studies with larger sample sizes and longer follow-up periods are recommended to provide more representative evidence regarding the burden and management of ACHD in Pakistan. Strengthening early screening, timely diagnosis, referral systems, and dedicated multidisciplinary ACHD services may also help improve long-term patient outcomes.

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