Original

# Patterns of Adult Congenital Heart Diseases at The Rivers State University Teaching Hospital

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## Abstract

**Background:** Adult Congenital heart defects (CHD) describe heart diseases present at birth. that persist into adulthood, and those corrected at childhood that have hemodynamic deficits requiring the attention of the adult cardiologists who manage complications such as pulmonary hypertension or surgical issues. There is paucity of studies on the Prevalence of Adult Congenital Heart Disease in Nigeria. This study aims to provide insights into adult congenital heart diseases (ACHD) at the Rivers State University Teaching Hospital

**Methods:** A retrospective review was conducted using data from the Rivers State University Teaching Hospital, Echocardiogram laboratory for a period of one year: from December 2022 to December 2023. The study involved 23 adult patients with confirmed CHD diagnoses. Data were collected from patient case notes and analyzed for demographic and clinical features and Echocardiogram diagnosis.

**Results:** The study included 23 adult patients with a mean age of 52.4 ± 22.1 years. The gender distribution was slightly female dominated (65.2%). CHDs were categorized into cyanotic and acyanotic types, with acyanotic defects being more prevalent (87.0%). The most common CHDs were atrial septal aneurysms (52.2%) and patent ductus arteriosus (13.0%). Cyanotic CHDs, such as Tetralogy of Fallot (TOF) and univentricular heart, accounted for 13.0% of patients.

**Conclusion:** The high prevalence of acyanotic CHDs and the significant occurrence of cyanotic CHDs highlight the need for specialized care and monitoring of ACHD patients in Nigeria. Improved health facilities, early diagnosis, and advanced interventional procedures are essential for managing ACHD effectively.

**Keywords:** Congenital heart defects, CHD; adult congenital heart diseases, ACHD; Nigeria; retrospective review, cyanotic CHDs, acyanotic atrial septal aneurysm, patent ductus arteriosus, Tetralogy of Fallot.



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## Introduction

Congenital heart defects (CHD) describe heart diseases present at birth. Many of these conditions are corrected early or result in early death, but some persist into adulthood. Adult cardiologists are often faced with managing complications arising from delayed interventions, such as pulmonary hypertension or surgical complications.[1]

Most adult congenital heart diseases may actually die in utero.[2] Autopsy studies in international African settings have shown discrepancies between clinical diagnoses and postmortem findings, highlighting the importance of accurate diagnosis and early intervention.[3] In Nigeria, autopsy findings have also revealed significant insights into the prevalence and types of congenital heart diseases.[4]

At birth, the prevalence of congenital heart diseases varies. Studies from Port Harcourt by Otaigbe et al.[5] have provided valuable data on the prevalence of congenital heart diseases among neonates. A study by Amaewhule et al[6] that recruited 530 neonates aged 0-7 days, showed congenital heart disease in 43 (8.1%) with a male to female ratio of 1.1:1. The commonest acyanotic defects were isolated atrial septal defect, patent ductus arteriosus, and ventricular septal defect, while the most common cyanotic defect was transposition of the great arteries. Other studies have also highlighted the common types of CHD in children, such as ventricular septal defects, patent ductus arteriosus, and atrial septal defects.[7-8]

Adult congenital heart diseases (ACHD) refer to congenital heart defects present at birth that persist into adulthood or treated congenital heart diseases with some hemodynamic abnormalities persisting into adulthood, requiring the attention of a cardiologist, specifically an adult cardiologist.[9] This field is a growing area of interest due to the increasing availability of advanced imaging tools and the globalization of medical knowledge through international conferences. As a result, there is a significant growth in personnel and manpower dedicated to this specialty.[10]

The prevalence of ACHD is increasing due to improved survival rates and better diagnostic techniques.[11] In the United States, it is estimated that about 1.4 million adults are living with congenital heart defects.[12] In Nigeria, the prevalence of ACHD is not well-documented, but A study suggest a significant number of adults present with conditions like shunt defects (ASD and PDA) for the first time.[13] In the Paediatrics population the late presentation of these conditions is often due to poor health facilities, inadequate screening modalities, and reliance on traditional birth attendants who may lack the skills to diagnose these defects. Even when diagnosed early, the high financial requirements for cardiothoracic procedures and the lack of skilled manpower for interventional procedures pose significant challenges. Nigeria is largely dependent on foreign missions to cover the care of only a limited number of patients.[14]

The field of ACHD is expected to see more growth in personnel and manpower as the demand for specialized care increases. International conferences and collaborations, such as those organized by the International Society for Adult Congenital Heart Disease (ISACHD), play a crucial role in advancing knowledge and training in this area.[15] These conferences provide a platform for healthcare professionals to share insights, discuss new research, and improve patient care.

In Nigeria, poor health facilities and inadequate screening modalities contribute to the late presentation of congenital heart diseases at birth resulting in missed diagnosis that persist only manifesting in adulthood. Many Nigerians resort to traditional birth attendants who are unskilled and may miss the diagnosis. Even when diagnosed early, the high financial requirement for cardiothoracic procedures and the lack of skilled manpower for interventional procedures pose significant challenges. Nigeria is largely dependent on foreign missions to cover the care of only a limited number of patients.[16]

There is a critical need locally for cardiologists who specialize in the care of adults with congenital heart diseases. These specialists are essential for managing complications arising from delayed interventions, such as pulmonary hypertension or surgical complications. As interventional procedures continue to improve, adult congenital heart disease clinics must adapt to meet the growing demand for specialized care.

In Nigeria, preliminary data from the National Paediatrics Cardiac Surgery Registry estimates that about 1,296 children are born with CHD annually.[17] A survey conducted by Chinawa JM et al.[12 -13] found that 71 out of 31,795 children evaluated in an outpatient clinic were detected with CHD over a 5-year period in a tertiary hospital in southeastern Nigeria. However, these hospital statistics may not be truly representative, as many births in Nigeria occur at home, unsupervised by a qualified doctor or midwife.

Globally, the incidence of CHD is approximately 8 in a thousand live births. The increasing prevalence of CHD globally, including in urban and rural areas of China, underscores the need for robust healthcare systems



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capable of providing timely and effective care for CHD patients.[14-15]

In Nigeria there is paucity of data on adult congenital heart diseases, this study seeks to provide information and stimulate interest, it also seeks to shed light on the pattern of congenital heart disease in Rivers State Nigeria.

## Methods

***Study Design:*** A retrospective review of data from an echocardiogram registry was conducted using data collected from a tertiary institution, the Rivers State University Teaching Hospital, over a period of one year (December 2022 to December 2023). The study design was cross-sectional.

***Setting:*** The study was conducted at the Echocardiogram laboratory of the Rivers State University Teaching Hospital, a tertiary institution in Rivers State, Nigeria. The relevant dates included the period of data collection from December 2022 to December 2023.

***Participants:*** The study population comprised all adult patients (18yrs and above) with suspected congenital heart disease who attended the clinic and had echocardiograms with a confirmed diagnosis of congenital heart disease. Consecutive adult patients diagnosed with CHD were recruited into the study. Confidentiality was maintained in handling patient data, and ethical clearance was sought and obtained from the Hospital's Ethical Committee.

***Variables:*** The variables included demographic data (age, gender), clinical features (tachypnea, dysmorphia, cyanosis, hypoxia, murmur), laboratory investigations, and diagnoses. Mean age, mean BMI, and blood pressures were obtained along with the clinical diagnosis.

***Data Sources/Measurement:*** Data were collected from Echocardiogram Register and entered into an Excel spreadsheet for analysis. Echocardiography were performed on all patients to confirm the diagnosis of congenital heart disease.

***Bias:*** Efforts were made to minimize bias by recruiting consecutive patients and ensuring accurate data entry. The study size was determined by the number of patients who attended the clinic during the study period.

***Study Size:*** The study included all adult patients with suspected congenital heart disease who attended the clinic and had echocardiograms with a confirmed diagnosis of congenital heart disease over the two-year period.

***Quantitative Variables:*** Quantitative variables such as age, BMI, and blood pressure were handled in the analyses. Groupings were chosen based on clinical type of Congenital Abnormalities.

***Statistical Methods:*** Statistical methods included descriptive statistics to summarize the demographic and clinical characteristics of the study population. Mean values and standard deviations were calculated for continuous variables, while frequencies and percentages were calculated for categorical variables. using the SPSS 25.

## Results

The study involved 23 adult patients with a mean age of 52.4 ± 22.1 years. With more females affected 14 (65.2%) of the study population. The mean BMI of the study population was 27.4 ± 5.3 kg/m², with mean systolic blood pressure of 115.3 ± 14.5 mmHg and mean diastolic BP of 73.6 ± 10.5 mmHg. Acyanotic CHDs was seen in 20 (86.95%) of study population, accounted for majority of the adult congenital heart disease with a wide spectrum ranging from atrial septal aneurysms (ASA), patent ductus arteriosus (PDA), bicuspid aortic valve, aortic right atrial fistula, and cotriatriatum. Cyanotic CHDs on the other hand accounted for only 3(13%) of patients, with Tetralogy of Fallot (TOF) and univentricular heart were the primary conditions; see table 1.

Clinically subjects with atrial septal aneurysm (ASA) presented with other cardiac abnormalities were symptomatic and in heart failure. However, two (2) had lone ASA (see table 2).

Patent Ductus Arteriosus (PDA) was found in three female patients, one was not in heart failure at the time of presentation but already had evidence of mild pulmonary Hypertension and was being prepped for surgical correction. The other two presented in Heart failure with Eisenmenger complex were managed and discharged to a cardiothoracic facility.

Atrial Septal Defect (ASD) was present in two patients: (1 male and 1 female), both with heart failure and Pulmonary hypertension. Other Conditions: Bicuspid aortic valve had periodic syncope; aortic right atrial fistula (had had post-surgical correction and was being cared for in pregnancy), The patient with cotriatriatum was asymptomatic while the subject with Pel Epstein



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anomaly presented in heart failure with massive ascites (see Figure 1).

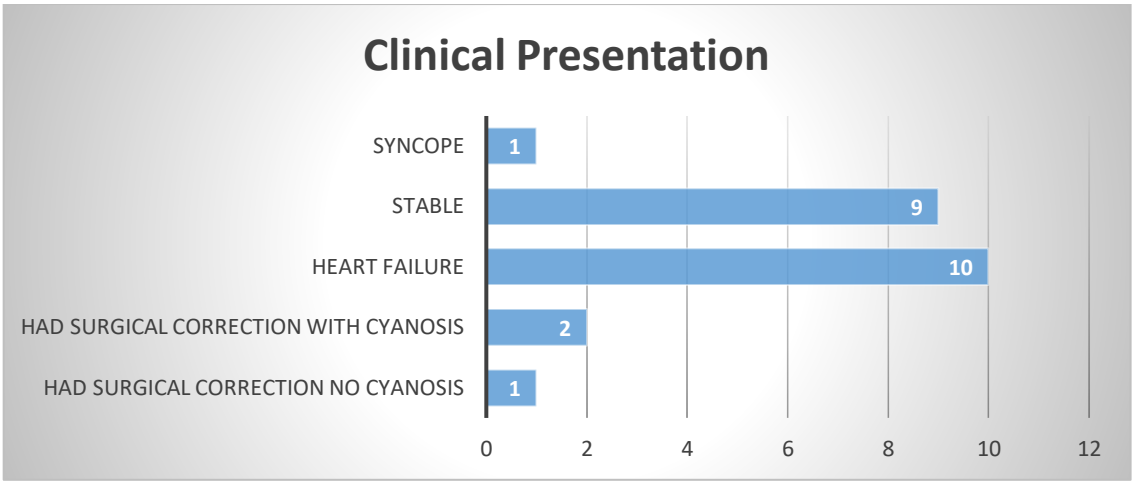


**Figure 1**. Clinical Presentation

Both Uni left Ventricles were very small for age and had central and peripheral cyanosis. One had polycythaemia of 60% and was being worked up for plasma exchange transfusion (PET). They had had some form of corrective surgery. One had Blalock Tausigs shunt insitu. Three (3) (13.04%)) of all the patients had had some form of corrective surgery whilst others were being prepped for surgery.

Figure 1 shows 3 Images, Image A: is a parasternal short axis view showing the interatrial septum with shunt defect consistent with an Ostium Secundum ASD. Image B: shows the origin of the tricuspid valve (TV) relative to the mitral valve (MV) with a long distance and Image C: A colour flow doppler interrogation of the tricuspid valve of the same patient with marked tricuspid regurgitation from the patient with Pel Epstein anomaly.

**Table 1**. Diagnosis Classification and Frequency

| | Diagnosis | Freq | Percent (%) |
|---|---|---|---|
| A | Acyanotic Congenital Heart Disease | 20 | 86.95% |
| B | Cyanotic Congenital Heart Disease | 3 | 13.04% |
| 1 | ASD with Pulmonary Hypertension/AS | 2 | 8.7% |
| 2 | All Atrial Septal Aneurysms | 11 | 47.83% |
| 1. | Atrial Septal Aneurysm | 2 | 8.7% |
| 2. | Atrial Septal Aneurysm AV Stenosis | 1 | 4.3% |
| 3 | Atrial Septal Aneurysm Desc Aorta Aneurysm | 1 | 4.3% |
| 4 | Atrial Septal Aneurysm Aortic Valve Sclerosis | 1 | 4.3% |
| 5 | Atrial Septal Aneurysm with HHDX | 2 | 8.7% |
| 6 | Atrial Septal Aneurysm (Thyrotoxic Heart: Type 2L) | 2 | 8.7% |
| 7 | Atrial Septal Aneurysm with COPD ASA | 1 | 4.3% |
| 8 | Atrial Septal Aneurysm with DCM IR | 1 | 4.3 |
| 3 | Bicuspid Aortic Valve | 1 | 4.3 |
| 4 | Patent Ductus Arteriosus | 3 | 13.0 |



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| | Diagnosis | Freq | Percent (%) |
|---|---|---|---|
| 5 | Pel Epstein Anomaly | 1 | 4.3 |
| 6 | Unileft Ventricle | 2 | 8.7 |
| 7 | Tetralogy of Fallot | 1 | 4.3 |
| 8 | Cotriatriatum | 1 | 4.3 |
| 9 | Aortico RA Fistula | 1 | 4.3 |
| | Total Number of Patients | 23 | 100 |

ASD: Atrial Septal Defect AV Stenosis: Atrioventricular Stenosis HHDX: Hypertensive Heart Disease Diagnosis COPD: Chronic Obstructive Pulmonary Disease ASA: Atrial Septal Aneurysm, DCM: Dilated Cardiomyopathy IR: Ischemic Reaction RA: Right Atrium

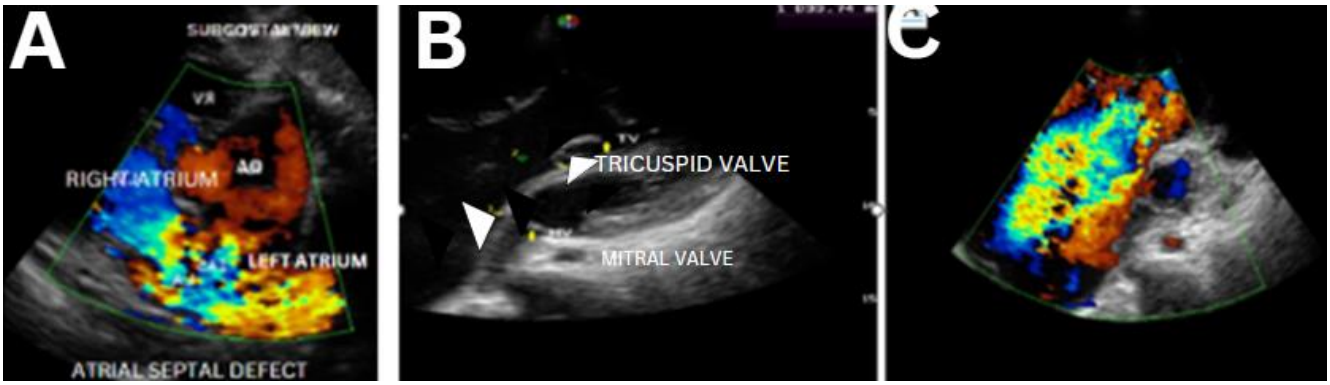


Figure 2: An Echocardiogram of an Ostium secundum type Atrial Septal Defect. B. Pel Epstein Anomaly. C. Color Flow showing severe TR Jets in Pel Epstein Anomaly.



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| Congenital Heart Disease | frequency |
|---|---|
| Atrial Septal Aneurysm | 12 |
| Uni left Ventricle | 2 |
| Atrial Septal Defect | 2 |
| Bicuspid Aortic Valve | 1 |
| PatentDuctus Arteriosus | 3 |
| Pel Ebstein Anomaly | 1 |
| Tetralogy of Fallot | 1 |
| Cotriatritum | 1 |
| Aortico Right Atrial Fistula | 1 |

**Figure 3:** Box Plot OF Body Mass Index and Diagnosis
With variability in BMI of patients with Atrial Septal Aneurysm and Patent Ductus arteriosus patients

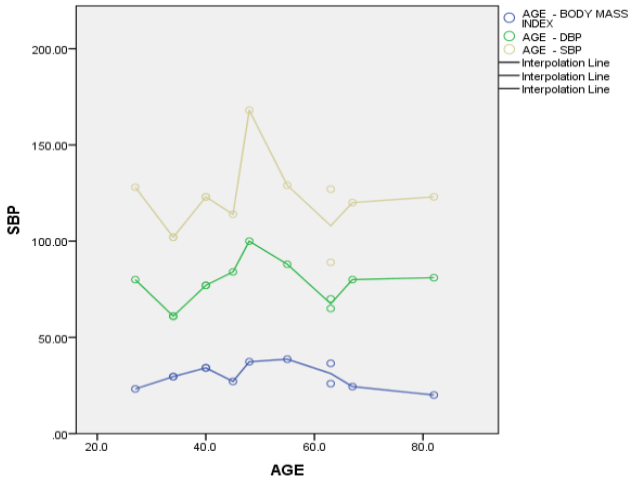


**Figure 4**: Scatter Plot representing the Relationship Between Age and BMI, SBP and DBP amongst Congenital Heart Disease Patients

## Discussion

The study included 23 adult patients diagnosed with various CHDs. The mean age of the patients was 52.4 ± 22.1 years, with a predominance of female patients (14 out of 23, 65.21%). This finding highlights the importance of targeted strategies for female patients, who may have distinct health needs and risk factors related to congenital heart diseases.

The mean age of 52.4 ± 22.1 years for this study population, rouses questions on the survival rates for adults with congenital heart disease (ACHD). This has improved significantly over the past decades, with advancements in medical and surgical treatments



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enabling over 90% of affected children to reach adulthood.[16] Despite this progress, adults with ACHD still face a higher risk of mortality compared to the general population, and more so in Nigeria when the financial burden of cardiothoracic surgical intervention is factored in. It is important to note that even milder conditions such as atrial septal defects carry elevated risks.[17] Studies reveal that at least 75% of patients with ACHD alive at 18 years of age live beyond middle age, with many reaching their sixties as noted in our study. Notably, those born after 1975 have experienced a substantial decline in mortality risks, reflecting improvements in care and management.[18-20] However, ACHD patients remain at higher risk of developing comorbidities such as atrial fibrillation, heart failure, and stroke, which contribute to the overall burden of disease.[21]

A predominance of females in the study population underscores the place of gender variations in congenital heart diseases (CHD). This point has been observed globally, with specific studies shedding light on these differences. For instance, a meta-analysis focusing on Down's syndrome patients revealed a higher prevalence of atrioventricular septal defects (AVSD) in females compared to males.[22] This suggests that certain genetic or biological factors may predispose females to specific types of CHD. Another study[23] highlighted that while males generally have a higher overall prevalence of CHD, females are more likely to present with severe forms of the disease, such as cyanotic CHDs. This disparity may be attributed to differences in genetic expression, hormonal influences, or even healthcare-seeking behaviors between genders. Additionally, a narrative review emphasized that gender differences extend beyond prevalence to include variations in clinical presentation and outcomes.[24] For example, females with CHD often experience worse outcomes post-surgery compared to males, potentially due to differences in body size, hormonal influences, or delayed diagnosis.[25]

The index study categorized congenital heart diseases (CHDs) into cyanotic and acyanotic types. Cyanotic CHDs, which include tetralogy of fallot (TOF) and uni-left ventricle, accounted for 13.0% of patients and are associated with higher risks of complications, necessitating specialized care. Acyanotic CHDs, making up 87.0% of cases, were further classified into shunt defects (atrial septal defect, patent ductus arteriosus, and aortico-right atrial fistula) and non-shunt defects (e.g., atrial septal aneurysm, bicuspid aortic valve, cotriatriatum, and Pel-Ebstein anomaly). The classification of CHDs in this study aligns with global categorization. Globally, cyanotic CHDs remain less common yet demand specialized care, while acyanotic CHDs often require long-term monitoring and management to address potential complications.[26]

Complementing this study, research in Enugu by Ejim et al[27] Nigeria, identified ventricular septal defects as the most common adult CHD, followed by atrial septal defects and Tetralogy of Fallot. The study by Ogundare et al[28] highlighted challenges in delivering cardiovascular services for CHD, emphasizing the need for government support and improved surgical care. Across Africa, a systematic review by Ndungu et al[29]. explored the prevalence and patterns of CHDs, while Mubangizi et al.[30] reviewed congenital and rheumatic heart diseases, highlighting the underestimation of the CHD burden due to limited access to care. These studies collectively underline the gaps in healthcare infrastructure and stress the importance of early detection, specialized care, and preventive strategies to improve outcomes for CHD patients across Nigeria and the continent.

The variability in clinical characteristics such as BMI, SBP, and DBP in the study population provide valuable insights into the overall health status of ACHD patients. These findings are consistent with global observations that emphasize the importance of individualized treatment plans based on patient-specific factors. The variability in BMI seen in the box plots for subjects with atrial septal defects may be explained by weight gain from ascites with pulmonary hypertension, resulting in right heart failure, this is also noted in the subjects with patent ductus arteriosus. Long-standing, uncorrected ASDs can lead to pulmonary hypertension, which in turn affects physical activity levels and overall metabolic rate, contributing to BMI variability. This also occurs in the patent ductus arteriosus (PDA) subject.[31]

The scatter plot analysis as seen in figure 4, revealed key insights into patients with congenital heart disease. A positive correlation was observed between systolic blood pressure (SBP) and age, indicating progressive vascular changes associated with the condition. Variations in Body Mass Index (BMI) across patients reflected differences in growth patterns, metabolism, and the presence of comorbidities. Outliers were noted, representing patients with significantly higher SBP or BMI values, warranting further investigation due to potential Comorbidities. Interpolation lines highlighted overarching trends, including the steady increase in SBP with age and the plateauing of BMI at certain age intervals. The variability in blood pressure and other clinical parameters highlights the complexity of managing ACHD and the fact that congenital heart



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disease can have other cardiovascular comorbidities as seen in patient with atrial septal aneurysm (table 2). There is therefore the need to individualize patient care and the need for continuous monitoring. Early detection and continuous monitoring are critical for improving outcomes in ACHD patients. Screening programs are important to identify undiagnosed CHDs in adults, enabling timely interventions

There is the need for specialized care and robust healthcare infrastructure to manage ACHD effectively. Globally, the establishment of dedicated ACHD clinics has been associated with improved patient outcomes.[32] These clinics provide a multidisciplinary approach to care, integrating, pediatric and adult cardiology, surgery, and patient education to address the complex needs of ACHD patients. Educating patients about their conditions and providing support for lifestyle modifications and adherence to treatment plans and encouraging research and collaboration among healthcare professionals to improve treatment outcomes and advance knowledge in the field of CHDs[33-34].

Only three (3) (13.04%) of all the patients had had some form of corrective surgery whilst others were being prepped for surgery, this speaks to the availability of funds, manpower and technology to care for the patients.

The prevalence of atrial septal aneurysm (ASA) varies significantly across studies, depending on the population and diagnostic methods used. In the general population, the prevalence of ASA is reported to range from 0.2% to 3%2.[35] However, in patients with cerebral ischemic events, the prevalence is notably higher. A study found that 7.9% of patients with cerebral ischemic events had ASA, compared to 2.2% in a control group.[36] Another study reported a prevalence of 27.7% in patients with cerebral ischemia and normal carotid arteries.[37]

The prevalence of atrial septal aneurysm (ASA) in patients with congenital heart disease varies across studies, reflecting differences in diagnostic approaches and population demographics. However, specific data on ASA prevalence within this group is less frequently reported. The index study at the Rivers State University Teaching Hospital stands out with a remarkably high prevalence of 47.83% for ASA among adults with congenital heart disease. Another study from Bangladesh: noted 111 patients with atrial septal aneurysm from and Echocardiographic review that enrolled 2598 over a one-year period.[38] This significant variation underscores the importance of standardized diagnostic criteria and methodologies to accurately assess ASA prevalence and its clinical implications in diverse populations.

ASA is clinically significant because it is often associated with patent foramen ovale (PFO), a condition that can facilitate paradoxical embolism. This mechanism contributes to cardioembolic strokes, particularly in younger patients with cryptogenic stroke. The presence of ASA increases the likelihood of PFO, and in many cases, ASA with or without PFO is the only potential source of embolism detected during echocardiography. Understanding the prevalence and implications of ASA is crucial for identifying at-risk individuals and implementing appropriate management strategies[39].

## Conclusion

This study highlights the significant burden of congenital heart diseases among adults in Nigeria, with a particular focus on the Rivers State University Teaching Hospital. The findings emphasize the necessity of early detection, continuous monitoring, and specialized care tailored to individual patient needs. A predominance of female patients, alongside variability in clinical characteristics, underscores the complexity of managing ACHD and the need for targeted strategies. Advancements in care have improved survival rates, yet ACHD patients continue to face elevated risks of mortality and comorbidities, particularly in resource-limited settings like Nigeria. To address these challenges, it is imperative to develop robust healthcare infrastructure, establish early screening programs, and foster multidisciplinary approaches to care. Additionally, educating patients, improving accessibility to surgical interventions, and encouraging research and collaboration are vital steps toward enhancing outcomes for ACHD patients. By bridging the gaps in healthcare delivery, policymakers and stakeholders can significantly improve the quality of life for individuals living with congenital heart diseases in Nigeria and beyond.

## Declarations

***Authors' Contribution:*** The lead author conceptualized the topic; other authors were involved in the funding and development. All authors read through and agreed on the final copy for submission.

***Conflict of interest:*** Authors declare no conflict of interest

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