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Pattern of Congenital Heart Diseases in Patients Presenting in a Tertiary Care Hospital of Karachi

Hajra Begum, Nadeem Sadiq, Saqlain Anwar, Ahmad Hassan, Zohab Ahmed, Hassan Waqar

ABSTRACT

Objective: To identify the frequency and pattern of congenital heart defects (CHD) in referred cases at a tertiary care hospital in Karachi, Pakistan.

Methodology: To carry out this research, we reviewed the medical records of referred patients from various regions of Sindh province from September 2023 to August 2024. The data was collected from the Pediatric Cardiology Department of a tertiary care hospital via a computerized data system and was analyzed to calculate the frequency and pattern of CHD.

Results: A total of 1,423 echocardiograms were reviewed; however, only 1,400 patients met the inclusion criteria and were included in the final analysis. Among these 1400 cases, 791(56.5%) were consistent with normal echo, and 609 (43.5%) were found to have a single or a combination of two or three CHDs. Among 609 CHD cases, 346 (56.8%) were male and 263 (43.1%) were female. Among these 609 cases, a total of 824 heart defects were diagnosed, 655 (79.4%) were acyanotic CHD, 119 (14.4%) were cyanotic CHD, and 50 (6.0%) others. In acyanotic heart defects, septal defects including VSD and ASD were most prevalent. However, among cyanotic heart defects, TOF was the most common abnormality identified.

Conclusion: CHD is very prevalent in Karachi and is among the most common causes of mortality and morbidity. Early detection and screening of heart defects through fetal and neonatal echocardiography allows timely interventions to improve this overall burden of mortality and morbidity.

Keywords: Atrial septal defect, Congenital heart defect, Tetralogy of Fallot, Ventricular septal defect,

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INTRODUCTION:

Congenital heart disease (CHD) consists of the most common birth defects in children worldwide with significant regional variability.[1] CHD has received considerable critical attention because of its potential to cause mortality in certain individuals and has become a growing public health concern worldwide.[2] As the word (congenital) suggests, these defects exist at birth and may persist throughout an individual's life, however often these defects resolve spontaneously, and the patient can lead their normal life without complications.[2-3] There are several types of congenital heart defects including malformations or abnormal development of some structures of the heart, the presence of abnormal openings within heart chambers, and even rhythm disturbances in some cases.[3] These defects are usually diagnosed in an individual's early life, but this early detection and screening depends upon the availability of adequate diagnostic equipment.[4] The importance of implementing systematic screening programs cannot be overstated, as they play a vital role in identifying CHD in asymptomatic patients.[5]

The birth defects often resolve spontaneously barely affecting an individual's health; however, if they persist they can potentially progress and increase the severity of the disease leading to complications and certain medical conditions including pulmonary hypertension, disrupting normal rhythms of the heart causing severe arrhythmia, certain infections to the heart structures and even congestive heart failure in severe cases.[6] The incidence of CHD is regionally dependent, but recent studies have concluded that this incidence is around 9.5/1000 live births globally[1] on average ranging from 4/1,000 to 50/1,000 live births.[7] and in Pakistan, 60000

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children are born with CHD annually with a prevalence of 3.4 per 1000 live births in the year 2023,[8] This statistic underscores that a significant segment of the Pakistani population is living with these birth defects, highlighting the necessity for improved healthcare strategies.[9]

The common types of congenital heart diseases (CHD) globally include Atrial Septal Defect (ASD), Ventricular Septal Defect (VSD), and Atrioventricular Septal Defect (AVSD). Other frequently observed conditions are Patent Ductus Arteriosus (PDA), Tetralogy of Fallot (TOF), Coarctation of the Aorta, Aortic Stenosis, and Pulmonary Stenosis. More complex forms of CHD include Tricuspid Atresia, Pulmonary Atresia, Transposition of the Great Arteries, and Single Ventricle anomalies. These conditions collectively represent a spectrum of structural heart abnormalities with varying clinical implications.[1,4]

It is now well established from a variety of studies, that, septal defects account for 40% of CHD in Pakistan, including ASD, VSD, and atrioventricular septal defects.[8] Early diagnosis of these defects plays a crucial role in controlling disease severity and timely interventions, in this way the outcome has been noticeably improved, enhancing the quality of life for individuals with these defects.[1] Moreover, targeted public health initiatives can facilitate access to specialized care and improve the overall management of CHD .[10]

The detection of congenital heart diseases (CHD) commonly involves a range of diagnostic tests. These include echocardiography, which provides detailed images of the heart's structure and function, and electrocardiography (ECG), used to evaluate the heart's electrical activity. Additional diagnostic tools include chest X-rays for assessing heart size and lung involvement, cardiac MRI and CT scans for advanced imaging and pulse oximetry to measure blood oxygen levels, which can indicate underlying cardiac issues. These tests play a crucial role in accurately diagnosing CHD and guiding appropriate management.[1]

In Pakistan, newborn screening for critical CHD involves a simple bedside test called pulse oximetry. Existing meta-analyses have found specificity for critical CHD of 99.9%, a sensitivity of about 76%, and a false positive rate of 0.14%.[1] The ultimate diagnosis of CHD comes via cardiac imaging, usually an echocardiogram.[1]

There is a lack of enough diagnostic facilities in certain regions of Pakistan[2] however, the past decade has seen increasingly rapid advances in the field of health and has established imperative means of early detection and screening of heart defects through fetal and neonatal echocardiography.[8] These diagnostic modalities are now under practice in most of the tertiary care pediatric hospitals nationwide.

METHODOLOGY:

This is a retrospective descriptive study in which 2D echocardiograms of pediatric patients referred to the cardiology department of PNS Shifa Hospital were analyzed. This study aimed to assess the trends of various congenital heart diseases and determine the frequency of cardiac abnormalities and distribution of different heart diseases based on gender and cause of referral for echocardiography.

We analyzed the echocardiograms of 1423 patients who were referred to PNS Shifa Hospital due to an abnormal clinical finding between September 2023 and August 2024. Only those patients who were selected were referred to our hospital due to an abnormal clinical finding that had not yet been diagnosed with congenital heart disease.

Investigators collected and analyzed 2D echocardiogram reports from hospitals' electronic health records. The data was categorized into several groups based on outcome (normal or abnormal), gender, type of disease, and reason for referral. The data was verified and cross-checked to prevent errors regarding patient age, address, and findings on the echocardiogram. The same pediatric cardiologist performed all echocardiograms of referred cases.

Descriptive statistical methods were used to analyze the data. The frequencies and percentages were calculated for each category including the distribution of normal and abnormal echocardiograms, types of diseases, gender distribution, and causes of referral for echocardiography.

RESULTS:

In our comprehensive evaluation involving a total of 1400 patients, we observed a significant prevalence of congenital heart disease (CHD). Among these patients, 791 individuals, representing 56.5%, were found to exhibit normal echocardiographic results. In contrast, 609 patients, constituting 43.5% of the evaluated cohort, were diagnosed with various forms of congenital heart disease, underscoring the importance of early detection and intervention in this population.

Gender Distribution: A closer examination of the 609 patients diagnosed with CHD revealed notable differences in gender distribution Table 1. The breakdown is as follows: Male patients: 346 cases, accounting for 56.8% of the diagnosed population, Female patients: 263 cases, representing 43.1%.

These figures indicate a distinct higher prevalence of congenital heart disease among male patients compared to their female counterparts. This observation aligns with existing literature suggesting that males are more frequently affected by CHD, potentially due to genetic or environmental factors. [11]

Distribution of Congenital Heart Diseases: 609 patients who presented with abnormal echocardiographic findings were diagnosed with a total of 824 cases of congenital heart disease. It is common for individuals to have multiple concurrent conditions, as reflected in our findings.

Among the 609 patients with confirmed CHD, a total of 824 distinct cardiac lesions were diagnosed, indicating that

multiple concurrent abnormalities were common. Acyanotic heart defects accounted for 79.4% of all anomalies, significantly outnumbering cyanotic conditions. Ventricular Septal Defect (VSD) was the most frequently observed lesion, representing 29.1% of all cases, followed by Atrial Septal Defect (ASD) and patent Ductus Arteriosus (PDA).

Cyanotic heart diseases made up 14.4% of all diagnoses. Tetralogy of Fallot (TOF) was the most common cyanotic lesion, accounting for 35.2% of cyanotic cases, followed by transposition of the great arteries (TGA), double outlet right ventricle (DORV), and pulmonary atresia. The higher frequency of TOF mirrors findings from previous Pakistani studies and suggests improved early detection strategies in tertiary referral centers. [3,14]

An additional 6% of diagnosed cases fell outside classical CHD classifications. These included dilated cardiomyopathy (DCM), bicuspid aortic valve (BAV), and left ventricular dysfunction. While not congenital in origin, these conditions were captured due to their significant structural or functional findings on echocardiography. DCM was the most common among these and, although classified as acquired or inherited rather than congenital, is an important differential diagnosis in symptomatic pediatric patients. [12]

Regarding referral patterns, heart murmurs were the most common reason for echocardiographic evaluation, accounting for 31.7% of referrals. Other prevalent indications included repeated upper respiratory tract infections (22.5%) and failure to thrive (9.7%). Prenatal detection accounted for 7.35% of referrals, illustrating the increasing utility of fetal echocardiography. A substantial proportion of referrals (21%) lacked documented causes, which may indicate inconsistent clinical documentation or generalized suspicion. These trends are supported by earlier literature recognizing murmur and respiratory symptoms as leading indicators of CHD. [13]

Overall, this study reaffirms known epidemiological patterns of CHD in South Asian pediatric populations, particularly the predominance of acyanotic lesions, the male preponderance, and the central role of physical exam findings in guiding diagnostic echocardiography. By comparing the frequency of defects such as VSD, ASD, PDA, and TOF with historical cohorts, this dataset contributes to the regional literature and underscores the importance of early referral and accessible cardiac imaging.[3,14,15,16]

DISCUSSION:

There are only limited studies carried out in Karachi regarding the understanding of the pattern and frequency of CHD, considering the persistent increase in the burden of these anomalies. Few studies were published but lacked the gender variation of these defects or the causes of referral were not discussed or mentioned in many articles. By conducting this study, we offer an understanding of the increase in prevalence and importance of clinical evaluation and examinations in patients presenting to resource-limited settings.[14-15] If properly evaluated these patients should be referred to the tertiary care settings which provide adequate diagnosis, *figure 1* thus ensuring the timely interventions and control of disease burden. In our study, even though many referred cases were not found to have cardiac anomalies (56.5%), (43.5%) cases were diagnosed with CHD. This shows the importance of a proper referral system which ensures better outcomes and adequate management of these diseases.

The present study provides valuable insights into the prevalence of various congenital heart diseases (CHD) compared to previous studies as indicated in Table 5.

The Ventricular Septal Defect (VSD) prevalence in our

Table 1: Echocardiographic findings of referred cases (n=1400), gender-wise distribution of congenital heart disease (CHD) patients (n=609), and classification of cardiac diseases (n=824)

2D ECHO OF REFERRED CASES	
Total	1400
Normal	791(56.5%)
Abnormal	609 (43.5%)
GENDER WISE DIVISION OF CHD PATIENTS	
Total	609
Male	346 (56.8%)
Female	263 (43.1%)
TYPES OF CARDIAC DISEASES	
Total	824
Acyanotic	655 (79.4%)
Cyanotic	119 (14.4%)
Other cardiac conditions (non-CHD)	50 (6.0%)

Table 2: Frequency of acyanotic heart diseases among the total cases

ACYANOTIC	655
Ventricular septal defect (VSD)	191 (29.1%)
Atrial septal defect (ASD)	157 (23.9%)
Patent ductus arteriosus (PDA)	142 (21.6%)
Pulmonary stenosis	56 (8.5%)
Coarctation of aorta (COA)	18 (2.7%)
Aortic regurgitation	15 (2.2%)
Partial anomalous pulmonary venous return (PAPVR)	15 (2.2%)
Mitral regurgitation	14 (2.1%)
AV septal defect (AVSD)	10 (1.5%)
Aortic stenosis	9 (1.3%)
Right ventricular outflow tract obstruction (RVOTO)	8 (1.2%)
Aortopulmonary window	7 (1.0%)
Anomalous origin of the right pulmonary artery (AORPA)	7 (1.0%)
Mitral stenosis	5 (0.7%)
Left ventricular outflow tract obstruction (LVOTO)	1 (0.5%)

study was found to be 29.1%, which is lower than the 32.1% reported in the study (Sadiq 2002)[16] and 40.6% in the study (Aman 2011),[18] but higher than the 21.5% in the study (Pate N 2016 Jan-Feb).[3] This variation may highlight differences in referral practices or diagnostic criteria across the studies.

Table 3 Frequency of cyanotic heart diseases and other cardiac conditions among the total cases

CYANOTIC	119
Tetralogy of Fallot (TOF)	42 (35.2%)
Transposition of great arteries (TGA)	21 (17.6%)
Double outlet right ventricle (DORV)	16 (13.4%)
Pulmonary atresia	15 (12.6%)
Tricuspid atresia	15 (12.6%)
Truncus arteriosus	4 (3.3%)
Single ventricle	3 (2.5%)
Bilateral superior vena cava (bilateral SVC)	1 (0.8%)
Total anomalous pulmonary venous return (TAPVR)	1 (0.8%)
Taussig-Bing anomaly	1 (0.8%)
OTHER CARDIAC CONDITIONS	50
(NON-CHD)	
Dilated cardiomyopathy (DCM)	30 (60%)
Bicuspid aortic valve (BAV)	12 (24%)
Dextrocardia	3 (6%)
Mild left ventricular dysfunction	3 (6%)
Hypertrophic cardiomyopathy (HCM)	1 (2.0%)

Table 4 Causes of referral

CAUSES OF REFFERAL	**Total (1400)**
Heart Murmur	445 (31.7%)
Repeated upper respiratory infections	315 (22.5%)
Failure to thrive	137 (9.7%)
Prenatal diagnosis	103 (7.35%)
Echo screening	95 (6.7%)
CXR abnormality	7 (0.5%)
Tachycardia/ Tachypnea	4 (0.2%)
Unknown	294 (21%)

For Atrial Septal Defect (ASD), our study showed a significantly higher occurrence of 23.9% compared to the 9.3% reported in the study (Pate N 2016 Jan-Feb).[3] This suggests an increasing recognition of ASD in our patient population, potentially reflecting improved awareness and referral systems in Karachi.

The prevalence of Patent Ductus Arteriosus (PDA) in this study was 21.6%, markedly higher than the 12.8% seen in the study (Aman 2011)[18] and 9.79% in the study (Ahmad 2002).[17] This finding might indicate an emerging trend or enhancements in diagnostic capabilities. In the case of Pulmonary Stenosis, the prevalence was 8.5%, which aligns closely with the study (Ahmad 2002)[17] reporting 7%, indicating consistency across these datasets.

Notably, Tetralogy of Fallot (TOF) was observed at a prevalence of 35.2% in our study, significantly higher than other studies. This finding underlines the necessity for heightened awareness and early diagnosis of cyanotic lesions in the clinical setting. Overall, our study confirms and expands upon the established trends in congenital heart disease (CHD), emphasizing the ongoing importance of conditions like VSD and TOF in pediatric cardiology practice

The study aimed to analyze the CHD trends among the referred cases to the pediatric cardiology department from different regions mainly Karachi, Pakistan. This included the gender distribution Figure 1, frequency of the diseases, and pattern of various types of congenital heart defects among the patients referred from other health institutes with

Figure 1: Gender wise distribution of common congenital heart defects

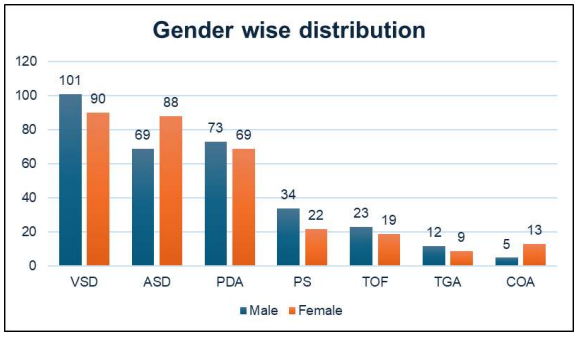

Table 5: Comparison of the frequency of common congenital heart diseases (VSD, ASD, PDA, PS, TOF) across different studies

Lesion / Study	**Sadiq (2002)[14] (n=6620)**	**Ahmad (2002)[15] (n=398)**	**Aman (2011)[16] (n=3072**	**Pate N (2016 Jan-Feb)[3] (n=1003)**	**Present study (n=609)**
VSD	1343 (32.1%)	180 (42.2%)	1248 (40.6%)	216 (21.5%)	191 (29.1%)
ASD	552 (13.2%)	56 (14.08%)	493 (16%)	93 (9.3%)	157 (23.9%)
PDA	536 (12.8%)	39 (9.79%)	394 (12.8%)	86 (8.6%)	142 (21.6%)
PS	336 (8.03%)	28 (7%)	236 (7.7%)	31 (3.1%)	56 (8.5%)
TOF	372 (16.1%)	38 (9.54%)	473 (15.4%)	245 (24.4%)	42 (35.2%)
No of patients	6620	398	3072	1003	609

suspicion of heart anomalies.

Due to limited diagnostic resources and equipment in basic health units across Pakistan, patients who show clinical signs suggestive of congenital heart defects (CHD) are typically referred to the nearest tertiary care facility for further assessment and management. As in our case there were around 1423 patients referred to PNS Shifa Hospital due to an abnormal clinic finding between September 2023 to August 2024 and 1400 patients were included in our study. The causes of these referrals are listed in the table. The trends observed in this study reveal a consistent pattern in the prevalence and types of congenital heart disease (CHD) encountered. It signifies that acyanotic heart diseases are more common, supported by previous literature which showed that out of 1003 studies 609 were acyanotic lesions and the most common acyanotic lesions were VSD[3] which is the case in our study as well. Among cyanotic lesions, we found the most common lesion was Tetralogy of Fallot as supported by previous literature.[19] Diseases that are not categorized as cyanotic or acyanotic heart defects are listed under the "others" category. Among these diseases Dilated Cardiomyopathy was found to be most prevalent in our study. However, existing studies have reported septal hypertrophy as the most prevalent.[3] This difference in findings is suggestive of some varying prevalence of heart anomalies across different ethnic groups.[20]

The data also indicates that heart murmur and upper respiratory tract infections are the most common causes of referral, which aligns with previous literature reporting heart murmur to be the most frequent symptom in patients with a cyanotic heart disease.[18] Our findings align well with existing literature, confirming the reliability of our data, and reinforcing the significance of our study in the broader landscape of congenital heart disease research. By supporting established trends, our study not only verifies these patterns but also emphasizes the ongoing importance of conditions like VSD and TOF in current clinical practice.[21] This alignment with previous research adds valuable context to our observations and enhances our understanding of the prevalence and distribution of CHD in our patient population.[22]

In conclusion, the necessity for increased awareness and better screening practices in our healthcare system is paramount.[23] Addressing the healthcare disparities that exist within Pakistan will facilitate early diagnosis and management of congenital heart diseases, ultimately reducing morbidity and mortality rates associated with these conditions.[24] Moreover, continuous training and development of healthcare professionals can improve the identification of CHD, particularly in primary care settings.[25] Finally, future studies should aim to explore the long-term outcomes of patients with CHD to provide comprehensive insights into the effectiveness of current treatment modalities.[26]

LIMITATIONS:

Our study has a few limitations as it was carried out in a single tertiary care facility and included only the patients who were being referred from resource-limiting settings. The results that were calculated might have suggested different frequencies and patterns of CHD if other centers of Pakistan were included and if the study were carried out on larger scale.

CONCLUSION:

CHD are among the most prevalent congenital diseases in Karachi and are among the most common causes of mortality and morbidity. We found that amongst 1400 cases that we reviewed VSD and TOF were the most common in acyanotic and cyanotic heart diseases respectively whilst the cause of referral was heart murmurs mostly. Early detection and screening of heart defects through fetal and neonatal echocardiography allows timely interventions to improve the overall burden of mortality and morbidity.

Authors Contribution:
Hajra Begum: Conceptualization of study design, writing
Nadeem Sadiq: Research supervision
Saqlain Anwar: Data analysis and interpretation
Ahmad Hassan: Literature search
Zohab Ahmed: Data collection and analysis
Hassan Waqar: Proof reading

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