RESEARCH ARTICLE

Frequency and pattern of adult congenital heart disease in a tertiary care cardiac hospital: Reasons associated with delayed diagnosis

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Abstract

Objective: To determine the distribution pattern and possible reasons of delayed diagnosis of congenital heart disease in adult population.

Method: The analytical, cross-sectional study was conducted at the Department of Paediatric Cardiology, National Institute of Cardiovascular Diseases, Karachi, from July to December 2021, and comprised patients of either gender aged 18 years and above with congenital heart disease confirmed by echocardiographic examination. Age, gender, weight, height, provincial affiliation, marital status, number of children, if any, duration of disease and New York Heart Association classification were noted on a predesigned proforma. Patterns of congenital heart disease types and reasons behind delayed diagnosis, if noted, were recorded. Data was analysed using SPSS 26.

Results: Of the 129 patients, 68(52.7%) were males. The overall mean age was 25.8±9.8 years. Mean age at the time of diagnosis was 12.4±14.8 years. There were 91(70.5%) cases with delayed diagnosis. There was no significant gender difference with respect to delayed diagnosis ($p>0.05$). Delayed referral 39(42.9%), delayed consultation 22(24.2%), delayed diagnosis by the doctor 21(23.1%), and social factors 9(9.9%) were the most frequent reasons behind delayed diagnosis. Ventricular septal defect was the most frequently defect 38(29.5%), followed by Tetralogy of Fallot 32(24.8%). Right ventricular dysfunction was the most common complication of delayed diagnosis 35(27.1%).

Conclusion: Delayed diagnosis of congenital heart disease was reported in over two-thirds of the cases, with ventricular septal defect being the most frequent type of defect, and delayed referral being the most common reason.

Keywords: Congenital heart disease, Diagnosis, Echocardiography, Tetralogy of fallot, Ventricular septal defect.

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Introduction

Congenital heart disease (CHD) refers to abnormalities in the structure, function or positioning of the heart that occur from birth, either manifesting early or remaining latent.[1] CHDs constitute 28-30% of all congenital abnormalities.[1,2] CHDs significantly impact infantile mortality rates, varying based on the type and severity of the condition.[3] In many parts of the world, inadequate healthcare and diagnostic facilities result in different occurrence rates among high- and low-income populations.

Recent advancements in the diagnosis and management of CHDs in children have significantly improved their prospects for a normal and productive life.[4-6] This has led to an increase in the number of CHD patients reaching adulthood above 18 years, forming a growing population known as "grown-up CHD patients (GUCHs)".[5] Despite higher treatment costs, the majority of GUCH cases require long-term medical care, contributing to an increasing burden on healthcare systems globally.[5] The adult CHD population is experiencing an annual growth of 5%, and in the United States alone, there is one adult with CHD among 1.3 million people.[7]

While regional hospital-based studies have been conducted to determine the prevalence of adult CHD (ACHD), national-level data reflecting the overall burden of CHDs in Pakistan, to our nest knowledge, is unavailable. Also, there is no systematic registry for ACHD cases, making it challenging to estimate the total number of affected individuals in the population. The lack of information about the age at which patients are initially diagnosed further complicates the picture. The current study was planned to determine the distribution pattern and possible reasons of delayed diagnosis of CHDs in adult population presenting to a tertiary care hospital.

Patients and Methods

The analytical, cross-sectional study was conducted at the Department of Paediatric Cardiology, National Institute of Cardiovascular Diseases (NICVD), Karachi, from July to December 2021. Approval was obtained from the institutional ethics review committee, and written informed consent was taken from all the participants. The

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sample was raised using convenience sampling technique. Those included were patients of either gender aged 18 years and above with CHD confirmed by echocardiographic examination. Newly-diagnosed as well as already diagnosed cases of CHD were included. Patients with unconfirmed diagnosis of CHD, a diagnosis of acquired heart disease, rheumatic heart disease, and patients who were unwilling to be part of this study were excluded.

During the study period, a total of 129 patients who fulfilled the inclusion and exclusion criteria, and presented to the department of cardiology were included. Data was collected on a predesigned proforma, and included age, gender, weight, height, provincial affiliation, marital status, number of children (if any), duration of disease, and the New York Heart Association (NYHA) classification.[5] Clinical data, including diagnosis, presenting signs and symptoms, and reason for possible delayed diagnosis or presentation, were noted. Reasons for consulting now, like symptoms, pregnancy and job requirements, were asked. Patterns of CHD types, severity, associated complications and management details were recorded.

Delayed diagnosis of cyanotic CHD was labelled as cases when a child with CHD was diagnosed after having been sent home from the birth clinic or hospital. Concerning acyanotic CHD, delayed diagnosis was defined as cases when the child was diagnosed at a stage when cardiac surgery or intervention should have already been performed.[8] In absolute terms, poverty was defined as earning <$2 per day.[9] An illiterate individual was characterised as someone who could not read and write a brief, simple statement about themselves in their daily routine.[10] Improper referral was defined as a referral which led to, or which could have led to, a different care pathway or worse clinical outcome than accepted standard practice.[11] Inadequate healthcare facilities were described as lacking the diagnostic tools, medications, equipment and skills required in a given healthcare facility to optimally manage a person with heart disease.[11] Social problems comprised distances between cultural or social factors, endangering the lives of social groups.[12]

Data was analysed using SPSS 26. Data was expressed as frequencies and percentages or as mean±standard deviation, as appropriate. Associations were estimated using chi-square test among qualitative data, while quantitative data was compared employing an independent sample t-test. P≤0.05 was taken as significant.

Results

Of the 129 patients, 68(52.7%) were males and 61(47.3%) were females. The overall mean age was 25.8±9.8 years, 44(34.1%) were married, and 97(75.2%) belonged to the Sindh province. The mean age at diagnosis was 12.4±14.8

Table-1: Gender-wise distribution of patients with respect to characteristics and CHD types.

Variables	Total	Gender		*p*-value
		Male	Female	
Total	n=129	n=68	n=61	
Mean Age (years)	25.8±9.8	24.7±8.7	27±10.8	0.179
Weight (kgs)	48.8±10.6	49.7±10	47.8±11.2	0.306
Height (cm)	165.8±9.9	168.1±9.2	163.3±10.1	0.005*
Marital Status				
Single	85 (65.9)	51 (75)	34 (55.7)	0.021*
Married	44 (34.1)	17 (25)	27 (44.3)	
No. of children	0.7±1.4	0.5±1.1	0.9±1.6	0.143
Province				
Sindh	97 (75.2)	51 (75)	46 (75.4)	0.340
Punjab	10 (7.8)	6 (8.8)	4 (6.6)	
KPK	7 (5.4)	4 (5.9)	3 (4.9)	
Baluchistan	11 (8.5)	7 (10.3)	4 (6.6)	
AJK	2 (1.6)	0 (0)	2 (3.3)	
GB	1 (0.8)	0 (0)	1 (1.6)	
Other	1 (0.8)	0 (0)	1 (1.6)	
Mean Age at first visit to NICVD (years)	16±14.8	13.8±13.1	18.4±16.3	0.080
NYHA classification				
I	14 (10.9)	7 (10.3)	7 (11.5)	0.273
II	87 (67.4)	45 (66.2)	42 (68.9)	
III	26 (20.2)	16 (23.5)	10 (16.4)	
IV	2 (1.6)	0 (0)	2 (3.3)	
CHD Types				
Septal defects/ DUCT				
Ventricular Septal Defect	38 (29.5%)	24 (35.3%)	14 (23%)	0.125
Atrial Septal Defect	32 (24.8)	12 (17.6)	20 (32.8)	0.047*
Patent Ductus Arteriosus	19 (14.7)	6 (8.8)	13 (21.3)	0.046*
Combination of simple defects	6 (4.7)	4 (5.9)	2 (3.3)	0.683
Obstructive defects				
Pulmonary stenosis	22 (17.1)	12 (17.6)	10 (16.4)	0.85
Coarctation of aorta	7 (5.4)	7 (10.3)	0 (0)	0.014*
Cong. mitral regurgitation	7 (5.4)	3 (4.4)	4 (6.6)	0.707
Tetrology of Fallot				
With spell	27 (20.9)	15 (22.1)	12 (19.7)	0.739
Without spell	6 (4.7)	3 (4.4)	3 (4.9)	0.892
CAVSD				
Balanced	4 (3.1)	3 (4.4)	1 (1.6)	0.621
Single ventricular heart (diagnosis)				
Inc. pul. Blood flow	3 (2.3)	2 (2.9)	1 (1.6)	0.624
Tricuspid atresia/ hypoplastic RV/ PS/ VSD	2 (1.6)	1 (1.5)	1 (1.6)	0.938
DTGA/VSD/PS	1 (0.8)	1 (1.5)	0 (0)	0.342
Dec. pul. Blood flow	4 (3.1)	3 (4.4)	1 (1.6)	0.621
Dextrocardia/ LTGA/PFO/PS	1 (0.8)	1 (1.5)	0 (0)	0.342
DORV/VSD/PS	1 (0.8)	1 (1.5)	0 (0)	0.342
TOF with hypoplastic LV	2 (1.6)	1 (1.5)	1 (1.6)	0.938
Trasnposition of great arteries				
DTGA with IVS	5 (3.9)	4 (5.9)	1 (1.6)	0.369
LTGA	2 (1.6)	1 (1.5)	1 (1.6)	0.938
DTGA with VSD	1 (0.8)	1 (1.5)	0 (0)	0.342
Total anomalous pul. Venous return				
Non obstructed	3 (2.3)	1 (1.5)	2 (3.3)	0.602
Bicuspid aortic valve	2 (1.6)	1 (1.5)	1 (1.6)	0.938
Cong. WPW synd.	1 (0.8)	0 (0)	1 (1.6)	0.473

CHD: Congenital heart disease, AJK: Azad Jammu Kashmir; CAVSD: Complete Atrioventricular Septal Defect; DORV: Double Outlet Right Ventricle; DTGA: D-Transposition of the Great Vessels; GB: Gilgit Baltistan; IVS: Interventricular septum; LTGA: Levo-Transposition of the Great Arteries; NICVD: National Institute of Cardiovascular Diseases; PFO: Patent Foramen Ovale; PS: Pulmonary Stenosis; RV: Right Ventricular; TOF: Tetralogy of Fallot; VSD: Ventricular Septal Defect.

Table-2: Gender-wise distribution of CHD cases with respect to reasons behind delayed diagnosis (n=79).

Variables	Total	Gender		*p*-value
		Male	Female	
Total	n=129 [n(%)]	n=68 [n(%)]	n=61 [n(%)]	-
Mean Age at diagnosis (years)	12.9±14.8	10.3±12.9	15.8±16.2	0.035
Delayed Diagnosis	91 (70.5)	47 (69.1)	44 (72.1)	0.708
Delayed referral	39 (42.9)	21 (44.7)	18 (40.9)	0.716
Delayed diagnosis	21 (23.1)	10 (21.3)	11 (25)	0.674
Delayed consultation	22 (24.2)	9 (19.1)	13 (29.5)	0.247
Social factors	9 (9.9)	6 (12.8)	3 (6.8)	0.342
Financial factors	14 (15.4)	9 (19.1)	5 (11.4)	0.304
Other	9 (9.9)	4 (8.5)	5 (11.4)	0.649

CHD: Congenital heart disease

Table-3: Frequency of complications (n=129).

Complications	n (%)
Inf. Endocarditis	2 (1.6)
Ventricular Septal Defect/ Pulmonary stenosis	1 (50)
Tetrology of Fallot	1 (50)
Arrhythmias	2 (1.6)
Atrial Septal Defect/ Cong. WPW synd.	1 (50)
Pulmonary stenosis	1 (50)
Heart blocks	1 (0.8)
Mixed septal defect: Ventricular Septal Defect/ Atrial Septal Defect	1 (100)
Eisenmenger syndrome	2 (1.6)
Atrial Septal Defect	1 (50)
Mixed septal defect: Ventricular Septal Defect/ Atrial Septal Defect	1 (50)
Ventricular dysfunction –BV	6 (4.7)
Atrial Septal Defect	1 (16.7)
Tetrology of Fallot	1 (16.7)
Bicuspid aortic valve	2 (33.3)
Single ventricular heart/ Dectrocardia/LTGA/PFO/PS	1 (16.7)
DTGA/VSD/Pulmonary Atresia	1 (16.7)
Ventricular dysfunction –LV	28 (21.7)
Ventricular Septal Defect	6 (21.4)
Atrial Septal Defect	1 (3.6)
Mixed septal defects	6 (21.4)
-Ventricular Septal Defect/Atrial Septal Defect	3 (10.7)
-Ventricular Septal Defect/Pulmonary stenosis	2 (7.1)
-Ventricular Septal Defect/Patent Ductus Arteriosus	1 (3.6)
LTGA	1 (3.6%)
Coarctation of aorta	5 (17.9)
Cong. mitral regurgitation	4 (14.3)
Pulmonary Atresia with VSD	1 (3.6)
Pulmonary stenosis	1 (3.6)
CAVSD Balanced	1 (3.6)
Single ventricle	3 (10.7)
-Pulmonary Atresia with VSD	1 (3.6)
-Tricuspid atresia/ hypoplastic RV/ASD/VSD/ PS	1 (3.6)
-CAVSD Balanced/DTGA/ Ventricular Septal Defect/Pulmonary stenosis	1 (3.6)
Ventricular dysfunction – RV	35 (27.1)
Tetrology of Fallot	19 (67.9)
Atrial Septal Defect	5 (17.9)
Atrial Septal Defect/Cong. WPW synd.	1 (3.6)
Ventricular Septal Defect	2 (7.1)
Mixed septal defect: Atrial Septal Defect/Patent Ductus Arteriosus	1 (3.6)
Pulmonary stenosis	5 (17.9)
Total anomalous pul. Venous return Non obstructed	1 (3.6)
Pulmonary Atresia with VSD	1 (3.6)

ASD: Atrial septal defect; CAVSD: Complete Atrioventricular Septal Defect; DORV: Double Outlet Right Ventricle; DTGA: D-Transposition of the Great Vessels; LTGA: Levo-Transposition of the Great Arteries; PFO: Patent Foramen Ovale; PS: Pulmonary Stenosis; RV: Right Ventricular; TOF: Tetralogy of Fallot; VSD: Ventricular Septal Defect, WPW: Wolff Parkinson White Syndrome, BV: Biventricular

years. Majority of the patients 87(67.4%) belonged to NYHA-II classification. Ventricular septal defect (VSD) was the most frequently observed CHD 38(29.5%), followed by tetralogy of fallot (TOF) 32(24.8%) (Table 1).

There were 91(70.5%) cases with delayed diagnosis. There was no significant gender difference with respect to delayed diagnosis ($p>0.05$). Delayed referral 39(42.9%), delayed consultation 22(24.2%), delayed diagnosis by the doctor 21(23.1%), and social factors 9(9.9%) were the most frequent reasons behind delayed diagnosis (Table 2).

Right ventricular (RV) dysfunction was the most common complication of delayed diagnosis 35(27.1%) (Table 3).

Discussion

In the present study, VSD and TOF were the most commonly noted CHD types observed in 29.5% and 24.8% of cases, respectively. A study published from the same centre in 2016 showed that TOF was the most frequent type of CHDs (24.4%), followed by VSD (21.5%) in all age groups. The current findings were in line with published local data is that VSD seems to be the most frequent CHD type in Pakistan, ranging from 21.5% to 46.0%, followed by TOF 15.4-25.3%.[13-15] A study in Lahore reported that 85.1% of CHD cases had delayed diagnosis.[16] Unlike Pakistan, in developed countries, neonatal clinical screening is performed for the detection of the majority of CHDs, and even foetal echocardiographic screening is used for the purpose.[17]

During early clinical screening, a considerable portion of CHD is overlooked and is identified after discharge from the hospital during childhood or even during adulthood.[18,19] A delay in diagnosis results in emergency situations.[20] The present study found that 70.5% cases had a delayed diagnosis of CHD. Meberg et al. found that of all those children who were initially considered healthy and discharged from the clinics, 24% were later diagnosed to have CHD.[21] According to Kuehl et al., 10% of all infants who die from CHD do so with only a postmortem diagnosis. They also found that a significant portion of infants with initially undiagnosed CHD experienced negative outcomes.[22] A delayed diagnosis in 10% of children was noted by Pfammatter et al., who required intervention for CHD, and due to this delay in referring them, 22% were reported to have complications.[20] Some researchers have documented GUCH patients with or without repair, underlining the fact that some adults with CHD are asymptomatic or have nonspecific physical examination findings, but some affected adults remain undiagnosed.[23] Additionally, there are not many studies on adults who have recently been diagnosed with CHD; they typically only include studies that report the incidence or

prevalence of adult CHD cases identified through clinical practice.[24]

The current study noted that delayed referral, delayed diagnosis by the doctor, delayed consultation and social structures were the most frequent reasons behind delayed diagnosis of CHD. A study found that delayed first consultation, delayed diagnosis by the doctor, and delayed referrals were the most common reasons behind delayed CHD diagnosis.[16]

In the current study, 66.7% adult CHD patients had comorbid cardiac complications. Murni et al. in India reported that in patients with delayed diagnosis of CHDs, 73.4% had comorbid complications.[8] A recent study in Brazil reported that 33.8% patients had complications, which is far lesser than what was observed in the current study.[25] Early detection and timely correction of the disease stand as pivotal measures to prevent complications, diminish mortality rates, and enhance the overall quality of life of individuals affected by CHDs.20 The unpredictability of complications associated with CHDs underscores their capacity to swiftly evolve into severe and potentially life-threatening conditions.[23]

The current study had limitations. It was a single-centre study at a referral centre situated in an urban setting. Multi-centre studies are needed, covering both urban and rural centres, to further explore the patterns of adult CHDs and the reasons behind delayed diagnosis. Being a single-centre study using a sample raised through convenience sampling technique limits and having potential selection bias has limited the generalisability of the current findings.

Despite the limitations, however, the growing population of adults with CHDs underscores the necessity for dedicated specialised centres tailored to their unique needs. With advancements in early diagnosis, surgical techniques, and interventions, a distinct subset of GUCH individuals has emerged. This evolving demographic requires specialised centres equipped to follow a multidisciplinary approach, and capable of providing comprehensive care that spans beyond traditional cardiology. These centres should not only address the complex medical needs, but also foster their integration as productive and responsible contributors to the national economy.

Limitations: The sample size was not calculated which can influence the generalizability of this research.

Conclusion

Delayed diagnosis of CHD was noted in over two-thirds of the cases, with VSD being the most frequent type of defect, delayed referral being the most common reason, and RV dysfunction being the most common complication of delayed diagnosis.

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Author Contribution:
AKA: Concept, design and collectively analysed the consent.
RS: Concept, design, data collection, drafting and collectively analysed the consent.
ASS: Data analysis and interpretation.
MM: Data analysis, interpretation and collectively analysed the consent.
SM: Data collection, drafting.
All authors operated the drafts.