Factors and Morbidities Associated with the Delayed Diagnosis of Congenital Heart Disease in Children Under 5

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Abstract

Objective: The aim of this study is to highlight the factors associated with the delayed diagnosis of Congenital Heart Disease and the resultant probability of developing life-threatening co-morbidities.

Method: A retrospective, population-based study was performed, consisting of 500 children with undiagnosed CHD, at Pediatric Medical Emergency, Children Hospital Faisalabad, from June 2021 to December 2021.

Result: According to the results, male predominance was observed (M: F = 1.3:1) with age ranging from 1 to 60 months. Most of them developed symptoms of CHD at or before the age of 1 month (n= 233; 47%) Only 62% were diagnosed at the age ranging between 2 to 6 months. Main factors for the delayed diagnosis of CHD were delayed first medical visit (n=230) and socioeconomic restraints (n=227). Main complications of CHD at the time of presentation were Hypovolemic shock (n=362; 72%) and Pulmonary Hypertensive Crisis (n=52; 10%).

Conclusion: The delayed diagnosis of CHD is associated with life-threatening complications and co-morbidities, which can be prevented with early diagnosis. The need of an hour is trained medical personnel, general public awareness, and easy access to specialized, well-equipped healthcare facilities.

Keywords: Congenital heart disease, pediatrics, morbidity, delayed diagnosis, complications

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Introduction

Congenital Heart Disease (CHD) is the most common congenital malformation worldwide.[1] In Pakistan, over 60,000 children are diagnosed with CHD every year.[2] However, the number of undiagnosed patients is much more. According to a study, 86 of 1000 Pakistani children with CHD die before they turn 5, about 44 die before the age of one month, accounting for 11% of total neonatal mortality rate.[3] The estimated prevalence of CHD is approximately 8 per 1000 live births.[3,4] Globally, the mortality rate of CHD patients has significantly declined and their quality of life is remarkably improved. But still, it accounts for a major portion of medical emergencies and pediatric health problems, worldwide.[1,4,5] By definition, CHD is structural or functional abnormality of the heart and/or the great blood vessels, that persists after birth.[6] There are over thirty-five types of CHD with variable clinical presentations. CHD is commonly divided into two sub-categories; This is a type of CHD where there is mixing of oxygenated and de-oxygenated blood.[6] The prominent presenting complaint is bluish discoloration of skin or Cyanosis due to low oxygen blood in the circulation, including Tetralogy of Fallot (TOF), Transposition of great vessels, Pulmonary Atresia, etc. It usually presents as medical emergency with obvious signs of CHD and cardiac de-compensation such as cyanosis, clubbing, respiratory distress, and congestive cardiac failure.[7,8] In this case, there will be no mixing of oxygenated and deoxygenated blood. The

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oxygen saturation in the blood will be adequate but the heart defect will result in the failure to supply the oxygenated blood efficiently throughout the body. It includes, Ventricular Septal Defect (VSD), Atrial Septal Defect (ASD), Patent Ductus Arteriosis (PDA), Aortic Valve Stenosis, Pulmonary Valve Stenosis, and Coarctation of Aorta.[7] Usually asymptomatic at the beginning but reveals itself at a certain age with the presenting complaints similar to that of lower respiratory tract infections, indigestion/neonatal colic, or asthma. These types of CHD can be easily misdiagnosed by general physicians, require expert evaluation followed by confirmative/diagnostic echocardiography.[6,7] Delayed diagnosis of congenital heart disease is prevalent globally whether it is high, middle, or low-income countries.[9] The accurate, early diagnosis of congenital heart disease can significantly control and manage the symptoms with decreasing the risks of long term complications and multi organ involvement.[10,11] Late diagnosis is associated with otherwise preventable morbidity, mortality, and disabilities. For example, most of the unattended or undiagnosed TOF patients later present with stroke, hemiparesis or hemiplegia. In that case, even if the CHD gets corrected, the associated morbidity will compromise the quality of life for a longer period of time or may be throughout the life.[11-14]

Patients with the suspicion of Congenital Heart Disease (previously un-diagnosed) confirmed with Echocardiography from CH & ICH, Cardiology Department Faisalabad.

Only those cases are included in the study where the attendants are the caregivers of the patient, to ensure the credibility of the collected data. Caregivers/Mothers not willing to participate in the study. Previously diagnosed cases of Congenital Heart Disease. This study aims to highlight the socioeconomic, perceptual, and behavioral factors that cause delay in CHD diagnosis and the resultant complications in the form of CHD associated morbidities. The results of this study can provide a basis for social education about the congenital heart diseases which may aid in its early diagnosis and prompt treatment. It also draws attention to the significance of prenatal screening to rule out CHD, which is currently not available to the women in Pakistan, especially in the rural areas.

Materials and Methods

The Concordat and the Outline of this study was approved by the Children Hospital Faisalabad's Ethical Review Board before the commencement of the study. To ensure the authenticity of the patient's history and collected data, only those children were included in the study where the attendants are the caregivers of the children. In the majority of the cases, the caregivers were mothers. After taking the verbal consent, detailed demographic history and relevant data was recorded. Statistical Analysis was performed using Statistical Package for Social Sciences Version 20 (SPSS-20).

Results

Over the period of six months, 500 undiagnosed CHD patients were enrolled in this study. Later on, the provisional diagnosis of CHD was confirmed by echocardiography. The results showed male predominance in CHD patients with male to female ratio 1.3:1, accounting for males n=299; Mean= 42.71; SD= 16.89 and females n=201; Mean= 28.71; SD= 11.68. Most of the patients presented at the age of 1 to 2 months comprising of 72 males (14.4%) and 55 females (11%). All the patients presented in pediatric medical emergency with the complications of congenital heart disease. In most of

Table 1: *Relationship between Age at the time of presentation at tertiary care hospital a frequency of confirmed cases of Congenital Heart Disease.*

Age Groups	Gender		Frequency	Valid Percent	Mean		Standard Deviation (SD)	
	Male	Female			Male	Female	Male	Female
1-2 Months	72	55	127	25.4%	42.7142857	28.7142857	16.89221533	11.68288474
2-6 Months	60	33	93	18.6%				
6-12 Months	46	21	67	13.4%				
12-24 Months	36	25	61	12.2%				
25-36 Months	38	27	65	13.0%				
37-48 Months	27	23	50	10.0%				
49-60 Months	20	17	37	7.4%				
Total	299	201	500	100.0%				

the cases, patients developed diarrhea associated with rapidly progressive, severe dehydration that precipitated the symptoms of CHD. Other documented complications of CHD were Cardiogenic Shock, Pulmonary Hypertensive Crisis, Infective Endocarditis, Recurrent Respiratory tract infections, Cerebrovascular Accident CVA.

Table.3 concludes all the possible factors that potentially caused delay in the diagnosis of Congenital Heart Disease in the selected patients. The most common factors among them were delayed first consultation with a doctor by guardians (n=230; 46%; p<0.001) and socioeconomic restraints (n=227; 45.4%; p<0.001).

Table 2: *Active complications of CHD documented at the time of their presentation*

	Active Complications	Frequency	Valid Percent
Due to delayed Diagnosis of CHD	Cardiogenic Shock	52	10%
	Recurrent RTI	12	2%
	Hypovolemic Shock due to severe dehydration	362	72%
	Pulmonary Hypertensive Crisis	52	10%
	CVA	3	1%
	Infective Endocarditis	19	4%

Table 3: *Frequency of the factors influenced the delayed diagnosis of CHD*

Possible Factors of Delayed Diagnosis of CHD	Present	Absent	Percentage
Socioeconomic Constraints	227	273	45.40%
Lack of Trained Health System	133	367	26.60%
Delayed First Consultation with a doctor by Guardians	230	270	46.00%
Delayed Diagnosis by Medical Professionals	124	376	24.80%
Delayed Referral to a Pediatric Cardiologist	118	382	23.60%
Social Taboos	212	288	42.40%
Most Children were delivered outside the Hospital	215	285	43.00%
Siblings more than 2	115	385	23.00%

Other include, delivery of children outside the hospital setup, social taboos, lack of trained health system, delayed diagnosis by medical health professionals, delayed referral to a cardiologist/ pediatric cardiologist, and more than 2 siblings.

Discussion

More than 80% of childbirths in rural areas occur at home by formal birth attendants (Daies).[15] Therefore, at the time of presentation, most of the mothers are unable to provide any documents regarding birth events of the child and/or unable to recall birth history.[16] Without proper history and expert's clinical examination, the signs and symptoms of CHD can be easily confused with respiratory tract infection.[17] Most of the children with congenital heart disease are misdiagnosed and mistreated in their early ages till they get access to tertiary healthcare center.[16-18] For cyanotic heart disease, delayed diagnosis is when an affected newborn is sent home after delivery without being diagnosed. For acyanotic heart disease, the delayed diagnosis is labeled when the patient is diagnosed after the age of elective surgical correction or after developing the hemodynamic instability as a complication of CHD.[19] All the patients included in this study were diagnosed at the time of their presentation in the Pediatric Medical Emergency, Children Hospital Faisalabad. Almost all types of congenital heart defects can be accurately diagnosed with pre-natal screening and in-utero fetal echocardiogram.[20] However, the misdiagnosis of CHD is still a major concern worldwide.[21] With the complex web of symptoms, vague history, co-morbidities, and lack of proper medical/ birth record makes it very difficult for a physician to diagnose CHD at the first visit. So the early diagnosis of CHD is a challenge for the primary healthcare physicians with their limited exposure, knowledge, and resources. Unfortunately, this relevant delay causes not only the morbidity and mortality of the patient but also the psychological and socioeconomic burden to the family.[22]

There are multiple factors that contribute to the delayed diagnosis of CHD. It mainly includes delay in the first consultation to a doctor by the guardians of the patient, socioeconomic restraints as people from rural areas have to travel far to access the specialized healthcare facility, and social taboos.[23] The social factors were defined as personal, cultural, and spiritual beliefs that hindered in seeking medical attention or being compliant with the CHD treatment.[21] These factors are more relevant to the acyanotic heart disease where patient is mostly asymptomatic or mildly symptomatic and do not develop obvious and more serious signs and symptoms such as peripheral and central cyanosis, apnea spells, etc. Other factors include lack of trained health system, delayed diagnosis by medical professionals, and delayed referral to a pediatric cardiologist.[22] These

delays transform into injudicious hospital admissions for symptomatic treatment resulting in the mismanagement of their medical condition as well as adding to the economic and psychological burden to their attendants or caregivers.[23-25]

The study shows that the patients with fewer siblings, 2 or less, had better chances to get access to the medical facilities and were diagnosed earlier than those with more than 2 siblings. This factor also justifies the socioeconomic restraints contributing to the limited resources and poor prognosis of affected children.[24-25]

Conclusion

Congenital Heart Disease is one of the most common congenital anomaly. Early diagnosis is the key factor to enable timely management of the disease and its associated complications. Unfortunately, the facility of prenatal screening is not available to the most of the pregnant females in developing countries like Pakistan. However, detailed history taking, careful clinical examination and timely referral to cardiologist for diagnostic echocardiography can remarkably control the disease and its adverse outcomes.

Conflict of Interest *None*

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ZA, MA: Writing of Manuscript