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ORIGINAL ARTICLE

Factors linked with the delayed diagnosis of congenital heart diseases. A single center experience from a tertiary care hospital of Pakistan.

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ABSTRACT… Objective: To note the factors linked with delayed diagnosis of congenital heart diseases (CHDs) among children. **Study Design:** Cross-sectional study. **Setting:** Department of Pediatric Cardiology, National Institute of Cardiovascular Diseases, Karachi, Pakistan. **Period:** September 2022 to February 2023. **Material & Methods:** We analyzed children aged up to 15 years who presented for the first time with echocardiographic evidence for the diagnosis of CHDs. Socio-demographic data along with clinical information like gender, age, residential status, maternal education status, history of maternal fetal echocardiography and the frequency of delayed diagnosis of CHDs were noted on a specially made proforma. Parent or guardians were asked for possible reasons of delayed diagnosis for CHDs. **Results:** In a total of 250 children, the mean age was 2.33±3.81 years while 162 (64.8%) children were aged below 2 years. There were 138 (55.2%) boys. Residential status of 159 (63.6%) children was rural while maternal educational status of 99 (39.6%) was illiterate. There were 164 (65.6%) children who were diagnosed to have cyanotic CHDs while 86 (34.4%) had acyanotic CHDs. Frequency of delayed diagnosis was noted among 182 (72.8%) children. In these 182 cases, delayed first consultation was the commonest factor behind delayed diagnosis noted in 78 (42.9%) children while missed diagnosis by the pediatrician was the 2nd most common factors linked with the delayed diagnosis of CHDs as described by 62 (34.1%) parents. **Conclusion:** Almost 3/4th of the studied children had delayed diagnosis of CHDs while cyanotic CHDs were the most common CHD types. Delayed first consultant and missed diagnosis by the pediatrician were the most common factors linked with delayed diagnosis of CHDs.

Key words: Acyanotic, Congenital Heart Disease, Cyanotic, Echocardiography, Illiterate.

INTRODUCTION

The contribution of congenital heart diseases (CHDs) to all types of major congenital deformities is substantial affecting 2 to 3% of newborns, whereas a varying prevalence (3 to 10 out of 1000 live births) occurs across the world.[1-4] In Pakistan, it is estimated that more than 40,000 newborns are born annually with CHDs.[5] An increasing incidence of CHDs has been shown in western data as etiological risk factors are escalating.[1]

Delayed diagnosis of CHDs is prevalent globally, whether it is high, middle, or low-income country.[6,7] An accurate, early diagnosis of CHDs can significantly control and manage the symptoms while decreasing the risks of long term complications and multiple organ involvement.[8] 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 CHDs gets corrected, the associated morbidity will compromise the quality of life for a longer period of time, or may be throughout the whole life.[9-11] Recent local data has shown the frequency of delayed diagnosis of CHDs to be 79.6% which warrants further research.[12]

The aim of the current study was to note the factors linked with delayed diagnosis of CHDs among children. The study results may not only enable healthcare providers to identify the factors causing the delay in diagnosis of CHDs but there

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were also thought to assist and plan awareness and screening programs to ensure timely diagnosis of CHDs in the pediatric population.

MATERIAL & METHODS

This was a cross-sectional study conducted at the department of pediatric cardiology, National Institute of Cardiovascular Diseases, Karachi, Pakistan from September 2022 to February 2023. Approval from Institutional Ethical Committee was taken (2021/1007, dated 28-10-2021) and informed consent was sought from parents/ guardians. Considering the frequency of delayed diagnosis of CHD as 79.6%[12] with 95% confidence level and 5% margin of error, the sample size was calculated to be 250.

We analyzed children aged up to 15 years who presented for the first time with echocardiographic evidence for the diagnosis of CHDs. Children with acquired heart disease or those having rheumatic heart disease were excluded. Socio-demographic data along with clinical information like gender, age, residential area, maternal education status, history of maternal fetal echocardiography and the frequency of delayed diagnosis of CHDs were noted on a specially made proforma. Parent or guardians were asked for possible reasons of delayed diagnosis for CHDs. Any child diagnosed with cyanotic CHDs but with having history of getting discharged from a birth clinic was labeled as delayed diagnosis. Child with acyanotic CHDs if presented at a time when an elective cardiac repair must have already been undertaken as per contemporary standards of paediatric cardiology were considered as delayed diagnosis.[13]

Data analysis was performed utilizing "Statistical Package for Social Sciences (SPSS)", version 26.0. Categorical data like gender, area of residence, literacy status, socio-economic status, types of CHDs, delayed diagnosis (yes/no) and reasons for delayed diagnosis were represented as frequency and percentages. Numeric data was given representation as mean and standard deviation.

RESULTS

In a total of 250 children, the mean age was 2.33±3.81 years while 162 (64.8%) children were aged below 2 years. There were 138 (55.2%) boys representing a boy to girl ratio of 1.2:1. Residential status of 159 (63.6%) children was rural while maternal educational status of 99 (39.6%) was illiterate. There were 164 (65.6%) children who were diagnosed to have cyanotic CHDs while 86 (34.4%) had acyanotic CHDs (Table-I).

Characteristics		Frequency (%)
Gender	Boy	138 (55.2%)
	Girl	112 (44.8%)
Age	<2	162 (64.8%)
	2-5	51 (20.4%)
	>5	37 (14.8%)
Residential status	Urban	91 (36.4%)
	Rural	159 (63.6%)
Maternal education	Literate	151 (60.4%)
	Illiterate	99 (39.6%)
Types of CHD	Cyanotic	164 (65.6%)
	Acyanotic	86 (34.4%)
Maternal fetal echocardiography done	Yes	19 (7.6%)
	No	231 (92.4%)

Table-I. Socio-demographic and clinical characteristics of studied children (n=250)

Frequency of delayed diagnosis was noted among 182 (72.8%) children as shown in Figure-1.

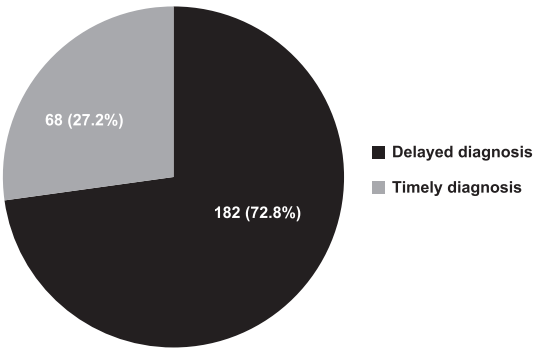

Figure-1. Frequency of delayed diagnosis of CHD (n=250)

Parents of the children with delayed diagnosis of CHDs (n=182) were further analyzed about the factors linked with delayed diagnosis of CHDs. It was found that delayed first consultation was the commonest factor behind delayed diagnosis noted in 78 (42.9%) while missed diagnosis by the pediatrician was the 2nd most common factors

linked with the delayed diagnosis of CHDs as described by 62 (34.1%) parents. Figure-2 is showing the frequency of factors for delayed diagnosis as described by the parents of the studied children.

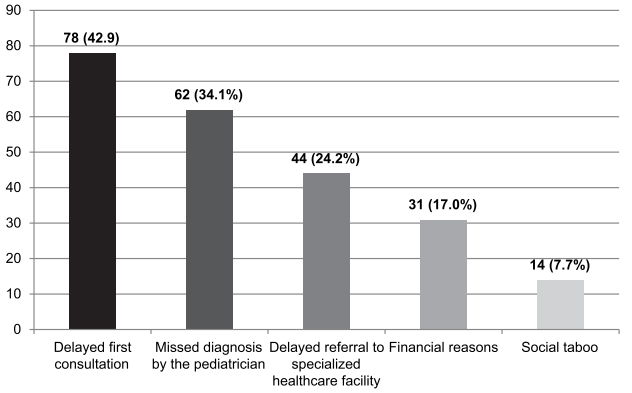

Figure-2. Frequency of factors described by parents for delayed diagnosis of CHDs (n=182)

DISCUSSION

It is revealed by our study that in 72.8% of children, there was a delay in the diagnosis of CHDs. Rashid U et al conducted a study in Lahore and described that delayed diagnosis of CHD was in 85% of children.[14] According to a Belgian study, in 9% of children, there was a delayed diagnosis of CHDs.[13] An American study on 3746 live-births with non-syndromic critical CHD showed that 29.5% of infants were detected late.[15] In both underdeveloped and developed countries, delayed diagnosis of CHDs has been documented and our study adds to what little is already known locally about it. Despite the fact that in the past few years, morbidity and mortality due to CHDs have significantly decreased, it is still recognized as an important health. Those patients who are in the process of cardiac interventions are likely to have cardiac compromise and organ dysfunction which can further result in the occurrence of prolonged ventilation and mortality if there is a delay in the diagnosis of CHDs.[10]

In the present study, 63.6% with CHDs were from rural areas. More than 80% of childbirths in rural areas occur at home through formal birth attendants (midwives).[15] Therefore, at the time of presentation, most of the mothers are unable to provide any record regarding the birth events of the child and/or are unable to recall the child's birth history.[16] Without a proper history and an expert's clinical examination, the signs and symptoms of CHDs can easily be confused with respiratory tract infections.[17] Most of the children with CHDs are misdiagnosed and mistreated in their early years, despite having access to tertiary health care centers.[16-18]

For cyanotic heart disease, it is considered a delayed diagnosis 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 hemodynamic instability as a complication of CHD.[19] All of the patients included in this study were diagnosed at the time of their presentation in the pediatric cardiac department. Almost all types of CHDs can be accurately diagnosed with pre-natal screening and an in-utero fetal echocardiogram.[20] However, the misdiagnosis of CHDs is still a major concern worldwide.[21] The complex web of symptoms, vague history, co-morbidities, and lack of a proper medical or birth record make it very difficult for a physician to diagnose CHDs at the first visit. So, the early diagnosis of CHDs is a challenge for 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 a psychological and socioeconomic burden on the family.[22]

There are multiple factors that contribute to the delayed diagnosis of CHD. It mainly includes delays in the first consultation with 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.[16,17] In this study, delayed first consultation was the commonest factor behind delayed diagnosis noted in 42.9% CHDs cases while missed diagnosis by the pediatrician was the 2nd most common factors linked with the delayed diagnosis of CHDs as described by 62 (34.1%) parents. Delayed first consultation has been pointed to be the most likely reason behind delayed diagnosis of CHDs

by other local researchers as well.[12,23] The social factors are described as personal, cultural, and spiritual beliefs that hindered seeking medical attention or being compliant with the CHD treatment.[12] These factors are more relevant to the acyanotic heart disease, where the patient is mostly asymptomatic or mildly symptomatic and does not develop obvious and more serious signs and symptoms such as peripheral and central cyanosis, apnea spells, etc. Other factors include a lack of a trained health system, delayed diagnosis by medical professionals, and delayed referral to a pediatric cardiologist.[24]

The present study hails from the biggest cardiac care center of our country so the findings of this study adds to important insights about the possible factors present behind delayed diagnosis of CHDs. Government and authorities may get some guidance from our findings about some of the important interventions aimed to assist timely diagnosis of CHDs. We were unable to prospectively follow the current set of children which is one of the limitations of this study. Further research studies involving long-term follow ups can be planned to record the impact of delayed diagnosis of CHDs among children.

CONCLUSION

Almost 3/4th of the studied children had delayed diagnosis of CHDs while cyanotic CHDs were the most common CHD types. Delayed first consultant and missed diagnosis by the pediatrician were the most common factors linked with delayed diagnosis of CHDs.

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3	Ram Chand	Proof reading, Data interpretation.	
4	Aliya Kemal Ahsan	Literature review, Methodology,	
5	Rajab Ali Khokhar	Data Collection, Final approval.	
6	Abdul Sattar Shaikh	Supervision, Critical revisions.	
7	Fazal ur Rehman	Data Collection, Drafting.