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Spectrum of Congenital Heart Disease in Paediatric Population Presenting to SKBZ Muzaffarabad

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Author`s Contribution

[1,5]Substantial contributions to the conception or design of the work; or the acquisition, [3,2,6]Active participation in active methodology, [4]analysis, or interpretation of data for the work, [6]Drafting the work or revising it critically for important intellectual content

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ABSTRACT

Objective: To determine the spectrum of congenital heart disease (CHD) in the pediatric population presenting to SKBZ Muzaffarabad.

Methodology: A descriptive cross-sectional, retrospective study was conducted in the Department of Pediatrics at SKBZ Muzaffarabad from March 2022 to February 2023. Children aged from birth to 5 years, of both genders, diagnosed with congenital heart diseases were included. After collecting demographic information and performing clinical assessments, patients were evaluated for congenital heart disease based on inclusion and exclusion criteria under the guidance of cardiologists. Additionally, relevant data from previous medical records were incorporated, as deemed sufficient for the study's analysis. All data were entered and analyzed using SPSS version 21.

Results: The overall mean age of the children was 10.87 months. The most common cyanotic defects were Tetralogy of Fallot (12.2%) and Transposition of the Great Arteries (4.1%). Acyanotic defects included Ventricular Septal Defect (VSD) (33.7%) and Patent Ductus Arteriosus (PDA) (12.2%). Other conditions, such as Dilated Cardiomyopathy (8.2%) and Dextrocardia with Situs Inversus (4.1%), were also observed, highlighting the diversity of CHD in the pediatric population. Tetralogy of Fallot (TOF) was more prevalent in boys, while Transposition of the Great Arteries was more frequent in girls. However, statistical analysis showed no significant gender-based variation across the various types of CHD ($p > 0.05$).

Conclusion: The study revealed a broad spectrum of congenital heart diseases among the pediatric population, with common cyanotic conditions including TOF and Transposition of the Great Arteries and common acyanotic defects such as VSD. Rare conditions like Dilated Cardiomyopathy and Dextrocardia with Situs Inversus were also observed.

Keywords: Congenital Heart Disease, Children, Cyanotic, Acyanotic.

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Introduction

Congenital heart disease (CHD) refers to a structural or functional abnormality of the cardiovascular system that is present at birth but is often identified later.[1] As the most prevalent major congenital anomaly, accounting for 28% of cases, CHD represents a significant global health challenge.[1] It fluctuates in degree of severity, with spontaneously returning interconnections between cavities to severe abnormalities that could require numerous procedures, including surgery or catheterization. It could lead to death during pregnancy, early childhood, or during the any age.[1] These disorders represent a significant category of pediatric illnesses and are a leading cause of childhood mortality and morbidity.[2] An estimated 8 cases of congenital heart disease occur per 1,000 live births, a figure widely accepted globally.[3] However, the prevalence varies across regions and countries due to genetic and environmental factors.[3,4] In Pakistan, statistics reveal that 86 out of every 1,000 children die before reaching the age of five, with 44 dying within their first month of life.[5] Among neonatal deaths, 11% are attributed to cardiac anomalies.[5,6]

Congenital heart disease (CHD) malformations can present as single defects or as combinations of multiple heart anomalies. Congenital heart disease (CHD) is commonly categorized into two types based on its pathophysiology and the specific heart structures involved: acyanotic CHD and cyanotic CHD.[7] Commonly identified isolated defects include atrial septal defects "ASD", pulmonary stenosis "PS" and the ventricular septal defects.[8] More complex anomalies, often involving multiple abnormalities, include tetralogy of Fallot, transposition of the great arteries and the atrioventricular septal defects.[8,9] Among these, TGA and the TOF are the most prevalent cyanotic CHD conditions, characterized by oxygen saturation levels below 90%. In contrast, septal defects like AVSD, VSD and the ASD are typically classified as acyanotic CHD, with oxygen saturation levels usually exceeding 95%.[8] Clinical

features and the severity depend on the different defects types, its impact on the hemodynamics, and the related anomalies. Clinical features may include respiratory distress, failure to thrive and respiratory infections recurrences, with certain defects being asymptomatic and only diagnosed later in the life. Accurate and the early detection in the critical to the optimize consequences, which may relies on the progression in the imaging modalities. Collaborative efforts across various fronts are actively addressing the global burden and inequities in pediatric cardiac care, aiming to achieve improved health outcomes for individuals regardless of their location or socioeconomic background.[10-12] In low income countries including Pakistan, numerous newborns and children die due to the lack of proper diagnosis and management for CHD. Delayed in diagnosis, inadequate specialized care and the financial difficulties make the situations worsening. However this study has been done to evaluate the most frequent congenital heart disease in the paediatrics population presenting at the SKBZ Muzaffarabad**.** Investigating the most common types of CHD among children is very important to understand the problem better, use resources wisely, and provide better care.

Methodology

This was a descriptive cross-sectional and retrospective study conducted in the Department of Pediatrics at SKBZ Muzaffarabad from March 2022 to February 2023. Children aged from birth to 5 years, of both genders, and diagnosed with congenital heart diseases (CHD) based on clinical evaluation and imaging studies—such as echocardiograms, ECGs, or chest X-rays—by pediatric cardiologists were included.

Children with acquired heart diseases, respiratory distress caused by non-cardiac factors (e.g., anemia and fluid overload, particularly in malnourished children), and those who did not consent to participate were excluded from the study.

The study was conducted after obtaining ethical approval from the Ethical Review Committee (ERC), and informed consent was obtained from each participant following thorough counseling. Participants were assured that their personal information would remain confidential and be used solely for research purposes. They were also informed that their data would contribute to identifying the most common congenital heart diseases in the population, which could aid in developing initiatives for early diagnosis and management.

Additionally, relevant data from previous records were incorporated when deemed sufficient for the study's analysis. After collecting demographic information and conducting clinical assessments, patients were evaluated for congenital heart diseases based on the inclusion and exclusion criteria under the guidance of cardiologists. All relevant information was entered and analyzed using SPSS version 21.

Results

This study included 49 children diagnosed with congenital heart disease. Among them, 26 (53.1%) were male, and 23 (46.9%) were female. The overall mean age of the participants was 10.87 months, with a standard deviation of 12.13 months (Table I).

Table I: Mean age and gender of the children with CHD (n=49)

Variables		**N**	**%**
Gender	Male	26	53.1%
	Female	23	46.9%
	Total	49	100.0%
Mean age (mean ±SD)		10.87±12.13 months	

In this study, the most common cyanotic defects were Tetralogy of Fallot (12.2%) and Transposition of the Great Arteries (4.1%). Among acyanotic defects, Ventricular Septal Defect (33.7%) and Patent Ductus Arteriosus (12.2%) were the most frequently observed. Additionally, other conditions such as Dilated Cardiomyopathy (8.2%) and Dextrocardia with Situs Inversus (4.1%) were also identified, highlighting the diverse spectrum of congenital heart disease (CHD) in the pediatric population, as shown in Table II.

Table II: Frequency of congenital heart disease (n=49)

Variables		N	%
Cyanotic	Tetralogy of fallout	6	12.2
	Transposition of great arteries	2	4.1
	Double outlet right ventricle	1	2.0
	Hypoplastic left heart	1	2.0
	Pulmonary atresia/stenosis	1	2.0
Acyanotic	Ventricular septal defect	16	33.7
	Atrial septal defect	3	6.1
	PDA	6	12.2
	AVSD	3	6.1
	VSD+ASD+PAD	1	2.1
	VSD and Mitral regurgitation/stenosis	1	2.0
	ASD+PAD	1	2.0
Others	Dilated cardiomyopathy	4	8.2
	Dextrocardia with situs inversus	2	4.1
	Hypertrophic cardiomyopathy	1	2.0
	Total	49	100.0

Tetralogy of Fallot was more common in boys, while Transposition of the Great Arteries was more frequent in

girls. Other conditions, including Ventricular Septal Defect (VSD), Atrial Septal Defect (ASD), and Dilated Cardiomyopathy, were distributed similarly between boys and girls. The analysis showed no significant gender-based variation across the various types of CHD, (p->0.05). Table III

Table III: Congenital heart disease according to gender of children (n=49)

Variables		GENDER		p-value
		Boys	Girls	
Cyanotic	Tetralogy of fallout	4(8.2%)	24.(1%)	0.346
	Transposition of great arteries	0(0.0%)	2(4.1%	
	Double outlet right ventricle	0(0.0%)	1(2.0%)	
	Hypoplastic left heart	1(2.0%)	0(0.0%)	
	Pulmonary atresia/stenosis	1(2.0%)	0(0.0%)	
Acyanotic	Ventricular septal defect	8(16.3%)	8(16.3%)	0.359
	Atrial septal defect	1(2.0%)	2(4.1%)	
	PDA	5(10.2%)	1(2.0%)	
	AVSD	1(2.0%)	2(4.1%)	
	VSD+ASD+PAD	0(0.0%)	1(2.0%)	
	VSD and Mitral regurgitation/stenosis	0(.0%)	1(2.0%)	
	ASD+PAD	1(2.0%)	0(0.0%)	
Others	Dilated cardiomyopathy	2(4.1%)	2(4.1%)	0.419
	Dextrocardia with situs inversus	2(4.1%	0(0.0%	
	Hypertrophic cardiomyopathy	1(2.0%	0(0.0%)	

Discussion

Congenital heart diseases (CHDs) in children are a significant contributor to infant morbidity and mortality. Early detection and timely management are crucial, as these conditions are often preventable.[13] However, this study was planned and conducted on 49 cases to determine the spectrum of CHDs in terms of the most common conditions among the pediatric population. The overall mean age of the participants was 10.87 months, with a gender distribution of 26 (53.1%) boys and 23 (46.9%) girls.

Consistently, Thomford NE et al.[8] reported a male-to-female ratio of approximately 1:1, with a median age of 1.9 years and an age range of 3 months to 16 years. Most of the patients in their study were under the age of 5 years, although their overall age range was broader compared to the present study. Similarly, Pasha W et al.[14] conducted a study involving 171 pediatric patients with CHD, reporting a mean age of 6.49 ± 3.56 months. Their study's gender distribution indicated a predominance of males (64.33%), with females accounting for 35.67%.

In contrast, Shah MA et al.[15] reported differing findings, with a relatively higher mean age of participants at 5.96 ± 1.954 years. In terms of gender distribution, males constituted 46.7% (176 individuals), while females were slightly more prevalent, accounting for 53.3%. These variations in mean age and gender proportions may reflect differences in study populations, sampling methods, or regional and demographic factors influencing the presentation and diagnosis of CHDs.

In this study, the most common cyanotic congenital heart defects (CHDs) were Tetralogy of Fallot (12.2%) and Transposition of the Great Arteries (4.1%), while among acyanotic defects, Ventricular Septal Defect (33.7%) and Patent Ductus Arteriosus (12.2%) were prevalent. Additionally, conditions such as Dilated Cardiomyopathy (8.2%) and Dextrocardia with Situs Inversus (4.1%) were also observed, emphasizing the diversity in CHD presentation in the pediatric population. When comparing these findings with other studies, Pasha W et al[14] reported that 74.26% of their patients had acyanotic CHDs, with VSD identified in 31.5%, ASD in 48.8%, a combination of VSD+ASD in 8.7%, and PDA in 11% of cases.

Cyanotic CHDs constituted 25.73%, with TOF being the most common (40.9%). Other cyanotic conditions included Transposition of the Great Arteries with VSD (15.9%), Tricuspid Atresia (29.5%), and Total Anomalous Pulmonary Venous Connection (TAPVC) in 13.6%. Similarly, Thomford NE et al[8] highlighted that acyanotic CHDs were the most frequently diagnosed, with VSD and PDA being the simplest and most common defects, while TOF was the most prevalent complex CHD, accounting for 25.5% of cases. Saif M et al[16] also noted that among 273 studied patients, 74.7% had acyanotic heart conditions and 25.3% had cyanotic heart diseases. VSD was the most frequently observed defect (29.6%), followed by TOF, which was present in 20.8% of the patients. Furthermore, Najaf Masood N et al[17] reported that 58.3% of their patients were diagnosed with acyanotic CHD and 41.7% with cyanotic CHD. Among the conditions, VSD was the most prevalent (33.3%), followed by TOF in 17.7%, making these the most common defects observed. Lastly, Khan MQ et al[18] found isolated VSD to be the most commonly observed acyanotic CHD (20.5%) and TOF as the most frequent cyanotic CHD, identified in 22.2% of the cases. Additionally few other studies also found comparable fisnidings.[19,20] These comparisons highlight the consistency in identifying VSD as the predominant acyanotic CHD and TOF as the leading cyanotic defect across different studies, reinforcing the need for early detection and management strategies tailored to the most

common CHDs.

Furthermore in this study, TOF was found to be more common in boys, whereas Transposition of the Great Arteries was more frequently observed in girls. Other congenital heart defects, such as VSD, ASD, and Dilated Cardiomyopathy, were distributed similarly between boys and girls. The analysis revealed no significant gender-based variation in the prevalence of the different types of congenital heart disease ($p > 0.05$). These findings are consistent with the study by Pasha W et al[14] which also reported no significant gender differences in the distribution of CHDs. Some variations in the frequency of congenital heart disease (CHD) were observed across different studies, which may be attributed to differences in study protocols, sample sizes, and study designs.

Geographic variations and differences in healthcare facilities, including access to specialized care and the frequency of medical visits by patients with CHD, may contribute to these observed discrepancies. However, this study has several limitations, such as a small sample size and the inclusion of retrospective data, which could affect the generalizability and accuracy of the findings.

To better understand the prevalence and characteristics of congenital heart disease (CHD) across different populations, further research involving larger, prospective cohorts and standardized methodologies is highly recommended. These studies could provide crucial insights to improve diagnostic accuracy and optimize treatment strategies for children affected by CHD.

Conclusion

This study revealed a range of congenital heart diseases among pediatric population, with common cyanotic conditions including Tetralogy of Fallot and Transposition of the Great Arteries, and acyanotic like as Ventricular Septal Defect. There were other certain other rarer conditions also observed. Early diagnosis and intervention are important for reducing preventable morbidity and mortality, particularly in regions with limited healthcare access. Based on the results and certain significant limitations of the study, further extensive studies are recommended to confirm the findings and provide a deeper exploration of these diseases and their contributing factors. This will help in developing health initiatives aimed at improving care for children with congenital heart disease.

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