



## A RETROSPECTIVE STUDY ON PATTERN OF CONGENITAL HEART DISEASE AMONG CHILDREN

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### ABSTRACT

**Background and Aim:** Congenital heart disease (CHD) is the most prevalent congenital anomaly among children, with over half of CHD-related deaths taking place during the neonatal period. The objective of the current investigation was to analyze the pattern of congenital heart disease among children.

**Patients and Methods:** A retrospective study was carried out on 254 CHD children in the Cardiology Department of Mardan Medical Complex, Mardan - Pakistan from August 2019 to August 2021. Demographic details of each patient included age, consanguinity, and gender were recorded. Other details such as pattern of CHD, presence of complaint and dysmorphic features, syndromes, and associated disorders were noted. Data analysis was done using SPSS version 27.

**Results:** Of the total 254 CHD children, there were 136 (53.5%) male and 118 (47.5%) female. Out of total cases, the incidence of acyanotic and cyanotic CHD was 78.7% (n=200) and 21.3% (n=54) respectively. The prevalent acyanotic congenital heart defect (CHD) was the isolated ventricular septal defect, accounting for 20.5% (n=41), whereas tetralogy of Fallot, at 22.2% (n=12), stood out as the most frequent cyanotic CHD. The most prevalent presentation was murmur found in 37% (n=94) patients. The incidence of heart failure, audible murmurs, maternal illnesses, consanguinity, prematurity, assisted reproduction, family history of CHD, abortions, and extra-cardiac anomalies are 43.7% (n=111), 72% (n=183), 51.6% (n=131), 46.5% (n=118), 18.9% (n=48), 10.2% (n=26), 8.7% (n=22), 7.1% (n=18), and 4.7% (n=12) respectively.

**Conclusion:** The present study found that accidental discovery of a murmur was the most common presentation among early-diagnosed children. Down syndrome is the most prevalent chromosomal disorder related to congenital heart disease (CHD).

**Keywords:** Congenital heart disease, Maternal disease, Cyanotic, Acyanotic, Consanguinity

## INTRODUCTION

Congenital heart disease (CHD) is the most prevalent congenital anomaly among children, with over half of CHD-related deaths taking place during the neonatal period [1]. The prevalence of CHD varies from 4 to 50 per 1,000 live births, as reported in studies conducted in different countries [2, 3]. Relying on the affected heart structure and pathophysiology, congenital heart disease is classified into cyanotic and acyanotic defects [4]. Ventricular septal defect (VSD), atrial septal defect (ASD), and atrioventricular canal defects are considered as mild CHD in acyanotic lesions. The complex acyanotic CHD consists of coarctation of the aorta and aortic stenosis [5]. Tetralogy of Fallot, hypoplastic left heart syndrome, tricuspid atresia, and transposition of great arteries are different conditions associated with cyanotic CHD [6]. Congenital heart disease's clinical presentation varies with age. Cyanosis, murmur, recent chest infection, Failure to thrive, and shortness of breath are different presentations of CHD [7]. Majority cases of CHD is caused by inheritance with multifactorial pattern including environmental and genetic factors but insignificantly associated with chromosomal aberrations [8, 9].

Congenital heart disease associated risk factors varies with regions around the globe [10]. A thorough examination of the epidemiology of congenital heart diseases (CHDs) serves as a crucial foundation for identifying the causes of cardiac dysmorphogenesis. This understanding is vital for creating opportunities to prevent these conditions prenatally [12, 13]. Unfortunately, there has been a lack of comprehensive studies on the epidemiology of CHD in Pakistani children. Therefore, the objective of the current investigation was to analyze the pattern of congenital heart disease among children.

## METHODOLOGY

A retrospective study was carried out on 254 CHD children in the Cardiology Department of Mardan Medical Complex, Mardan - Pakistan from August 2019 to August 2021. Demographic details of each patient included age, consanguinity, and gender were recorded. Other details such as pattern of CHD, presence of complaint and dysmorphic features, syndromes, and associated disorders were noted. Cyanosis, murmur, recent chest infection, Failure to thrive, and shortness of breath are different presentations of CHD recorded. Other details included tetralogy of fallot, hypoplastic left heart syndrome, tricuspid atresia, and transposition of great arteries are different conditions associated with cyanotic CHD. Data collection, tabulation, and analysis were conducted using SPSS, version 27. Numerical parameters were expressed as mean and standard deviation whereas categorical variables were described as frequency and percentages.

## RESULTS

Of the total 254 CHD children, there were 136 (53.5%) male and 118 (47.5%) female. Out of total cases, the incidence of acyanotic and cyanotic CHD was 78.7% (n=200) and 54 (21.3%) respectively. The prevalent acyanotic congenital heart defect (CHD) was the isolated ventricular septal defect, accounting for 20.5% (n=41), whereas tetralogy of Fallot, at 22.2% (n=12), stood out as the most frequent cyanotic CHD. The most prevalent presentation was murmur found in 37% (n=94) patients. The incidence of heart failure, audible murmurs, maternal illnesses, consanguinity, prematurity, assisted reproduction, family history of CHD, abortions, and extra-cardiac anomalies are 43.7% (n=111), 72% (n=183), 51.6% (n=131), 46.5% (n=118), 18.9% (n=48), 10.2% (n=26), 8.7% (n=22), 7.1% (n=18), and 4.7% (n=12) respectively. Age-wise distribution of patients are shown in Table-I. Frequency of different types of CHD are depicted in Figure-1. Mode of presentation of studied population are shown in Table-II. Incidence of different risk factors are demonstrated in Figure-2.

**Table-1** Age-wise distribution of children (N=254)

Age Group	N (%)
Neonatal	82 (32.3%)
<1 year	120 (47.2%)
1-5 years	28 (11.0%)
6-10 years	24 (9.4%)
<b>Total N (%)</b>	<b>254 (100%)</b>

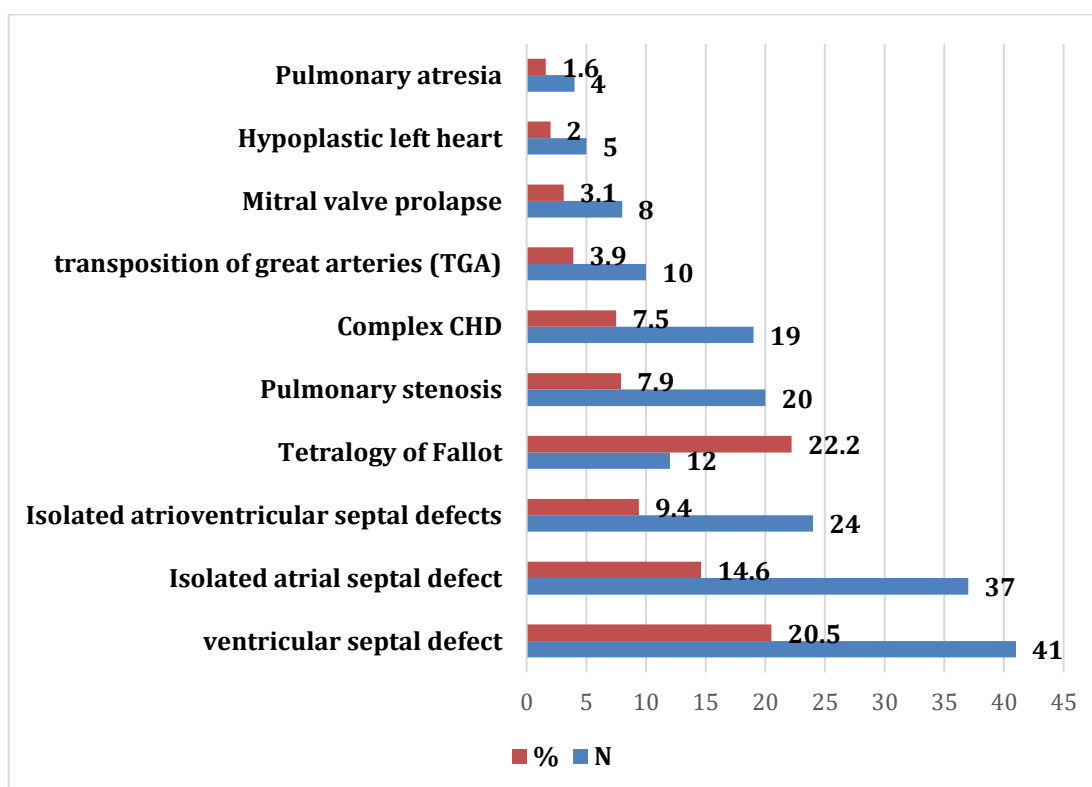


Figure-1 Frequency of different CHD (N=254)

Table-II Mode of presentation of studied population (N=254)

Presentation	N (%)
Murmur	94 (37%)
Recurrent chest infections	76 (29.9%)
Cyanosis	40 (15.7%)
Failure to thrive	32 (12.6%)
Neonatal sepsis-like disease	9 (3.5%)
Shortness of breath	3 (1.2%)
<b>Total N (%)</b>	<b>254 (100%)</b>

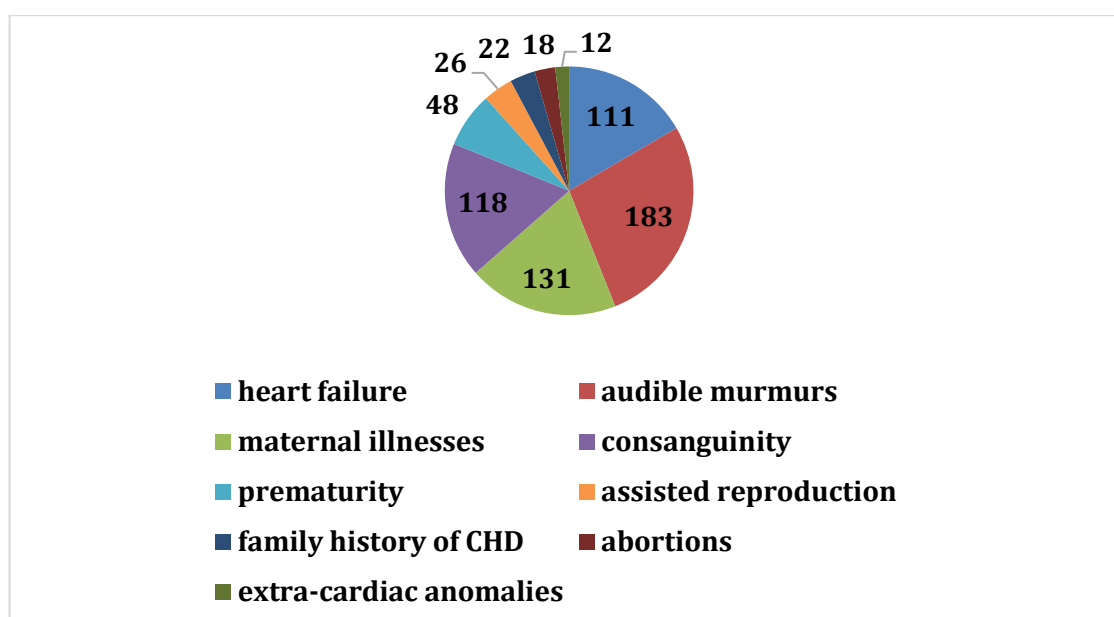


Figure-2 Incidence of different risk factors (N=254)

## DISCUSSION

The clinical presentation of congenital heart disease (CHD) exhibits variability and is influenced by the age of the individual, necessitating a heightened level of suspicion for diagnosis and intervention at early stages [14]. The accidental discovery of murmur, cyanosis, neonatal sepsis-like illness, recurrent chest infections and shortness of breath were the prominent CHD presentation. Al-Ammouri et al. [15] reported that the incidence of different indications on echocardiography such as murmur, weight gain failure, rapid breathing, and cyanosis was 35%, 10%, 20%, and 10% respectively. Similarly, Khasawneh et al. [16] observed that weight gain failure and rapid breathing were the major presentation of children suffering from congenital heart disease.

Tetralogy of Fallot continues to stand out as the most prevalent cyanotic heart defect, consistent with findings reported in other studies [17-19]. Notably, there exists a sizable population of unrepaired patients who are still alive, indicating enhanced survival rates, particularly in less severe cases. The rising incidence of congenital heart disease (CHD) can be attributed, in part, to the increased antenatal detection of suspected cases, leading to early referrals for cardiac evaluation. Antenatal identification plays a crucial role in preventing delayed diagnoses and mitigating associated consequences [20]. Furthermore, early detection during pregnancy is anticipated to enhance operative outcomes and improve the prognosis.

The occurrence of complex and rare types of congenital heart defects (CHDs) was found to be lower compared to Western data but similar to findings in other Indian studies [21, 22]. This difference could be attributed to the severity of the defects, potentially leading to patient mortality before accessing medical facilities, as well as variations in racial and genetic factors between different populations. The current prevalence of CHDs is lower than in an earlier study from the same region, possibly due to improved peripheral health services. It is likely that certain cases of CHD went undiagnosed, especially among neonates born at home who may have passed away without medical attention. Additionally, asymptomatic cases with mild to moderate CHD or those diagnosed at peripheral or private healthcare centers may contribute to a falsely low prevalence. It is noteworthy that the diagnosis of CHD can go unnoticed in up to 30% of infants during the first weeks of life [23]. Congenital heart disease (CHD) stands out as the predominant cause of heart failure. An earlier study reported that 45% children had heart failure [24]. Another study concluded that 40% children had heart failure among CHD cases [25]. The increasing rate of complications may be related to factors such as irregular follow-up and noncompliance with treatment, potentially resulting in delayed surgical management. Earlier studies [26, 27] investigated the association of incidence of CHD with consanguinity and found that consanguinity was in 44.6% and a positive CHD family history in 9.2% of the studied population, which is comparable to our result and with the findings of Zhao et al. [28], who reported consanguinity in 49% and 14% a positive family history.

## CONCLUSION

Accidental discovery of a murmur was the most common presentation among early-diagnosed children. Down syndrome is the most prevalent chromosomal disorder related to congenital heart disease (CHD).

## REFERENCES

1. Namuyonga J, Lubega S, Aliku T, Omagino J, Sable C, Lwabi P. Pattern of congenital heart disease among children presenting to the Uganda Heart Institute, Mulago Hospital: a 7-year review. *African health sciences*. 2020 Jul 22;20(2):745-52.
2. Zahid SB, Jan AZ, Ahmed S, Achakzai H. Spectrum of congenital heart disease in children admitted for cardiac surgery at Rehman Medical Institute, Peshawar, Pakistan. *Pakistan journal of medical sciences*. 2013;29(1):173.doi:10.12669/pjms.291.2910.
3. Johar D, Ahmed SM, El Hayek S et al (2019) Diabetes-induced proteome changes throughout development. *Endocr Metab Immune Disord Drug Targets* 19:732–743. <https://doi.org/10.2174/1871530319666190305153810>.

4. Abushouk AI, El-Husseny MWA, Bahbah EI et al (2017) Peroxisome proliferator-activated receptors as therapeutic targets for heart failure. *Biomed Pharmacother* 95:692–700. <https://doi.org/10.1016/j.biopha.2017.08.083>
5. Abqari S, Gupta A, Shahab T et al (2016) Profile and risk factors for congenital heart defects: a study in a tertiary care hospital. *Ann Pediatr Cardiol* 9:216. <https://doi.org/10.4103/0974-2069.189119>.
6. Majeed-Saidan MA, Atiyah M, Ammari A, Alhashem A, Maha S, Mohamed M, et al. Patterns, prevalence, risk factors, and survival of newborns with congenital heart defects in a Saudi population: a three-year, cohort casecontrol study. *J Congenit Cardiol.* (2019) 3. doi: 10.1186/s40949-019-0023-8.
7. Narayen IC, Blom NA, Ewer AK, Vento M, Manzoni P, Te Pas AB. Aspects of pulse oximetry screening for critical congenital heart defects: when, how and why? *Arch Dis Child Fetal Neonatal Ed.* (2016) 101:F162–7. doi: 10.1136/archdischild-2015-309205.
8. Trevisan P, Zen TD, Rosa RFM, da Silva JN, Koshiyama DB, Paskulin GA. Chromosomal Abnormalities in Patients with Congenital Heart Disease. (2018). Available online at: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4106807/> (accessed January 20, 2020).
9. Best KE, Rankin J (2016) Is advanced maternal age a risk factor for congenital heart disease? *Birth Defects Res A Clin Mol Teratol* 106:461–467. <https://doi.org/10.1002/bdra.23507>.
10. Wann KA, Shahzad N, Ashraf M, Ahmed K, Jan M, Rasool S. Prevalence and spectrum of congenital heart diseases in children. *Heart India.* 2014;2(3):76. doi:10.4103/2321-449X.140230.
11. Zaqout M, Aslem ES, Oweida FS, De Wolf D. Prevalence of congenital heart disease among Palestinian children born in the Gaza Strip. *Cardiology in the Young.* 2014;24(5):905-9. doi:10.1017/S1047951113001418.
12. Tankeu AT, Bigna JJ, Nansseu JR, Aminde LN, Danwang C, Temgoua MN, Noubiap JJ: Prevalence and patterns of congenital heart diseases in Africa: a systematic review and meta-analysis protocol. *BMJ Open.* 2017, 7:e015633. 10.1136/bmjopen-2016-015633
13. Yeh SJ, Chen HC, Lu CW, et al.: Prevalence, mortality, and the disease burden of pediatric congenital heart disease in Taiwan. *Pediatr Neonatol.* 2013, 54:113-8. 10.1016/j.pedneo.2012.11.010.
14. Saxena A, Mehta A, Sharma M, Salhan S, Kalaivani M, Ramakrishnan S, Juneja R: Birth prevalence of congenital heart disease: a cross-sectional observational study from North India. *Ann Pediatr Cardiol.* 2016, 9:205-9. 10.4103/0974-2069.189122.
15. Al-Ammouri IA, Ayoub F, Tutunji L. (PDF) Incidence of Congenital Heart Disease in Jordanian Children Born at Jordan University Hospital; a Seven-Year Retrospective Study. (2020). Available online at: <https://www.researchgate.net/publication/321136989>.
16. Khasawneh W, Hakim F, Abu Ras O, Hejazi Y, Abu-Aqoulah A. Incidence and Patterns of Congenital Heart Disease Among Jordanian Infants, a Cohort Study From a University Tertiary Center. *Frontiers in pediatrics.* 2020;8:219. doi:10.3389/fped.2020.00219.
17. Ou Y, Mai J, Zhuang J, Liu X, Wu Y, Gao X, et al. Risk factors of different congenital heart defects in Guangdong, China. *Pediatric research.* 2016;79(4):549-58. doi:10.1038/pr.2015.264.
18. Wann KA, Shahzad N, Ashraf M, Ahmed K, Jan M, Rasool S. Prevalence and spectrum of congenital heart diseases in children. *Heart India.* 2014;2(3):76. doi:10.4103/2321-449X.140230.
19. Zaqout M, Aslem ES, Oweida FS, De Wolf D. Prevalence of congenital heart disease among Palestinian children born in the Gaza Strip. *Cardiology in the Young.* 2014;24(5):905-9. doi:10.1017/S1047951113001418.
20. Derakhshan R, Barfeii N, Sadeghi T. Frequency of Congenital Heart Disease in Neonates with Extra Cardiac Anomalies. *International Journal of Pediatrics-Mashhad.* 2020;8(7):11597-602.
21. Chou H-H, Chiou M-J, Liang F-W, Chen L-H, Lu T-H, Li C-Y. Association of maternal chronic disease with risk of congenital heart disease in offspring. *Cmaj.* 2016;188(17-18):E438-E46. doi:10.1503/cmaj.160061.

22. Stoll C, Dott B, Alembik Y, Roth MP. Associated noncardiac congenital anomalies among cases with congenital heart defects. *Eur J Med Genet.* (2015) 58:75–85. doi: 10.1016/j.ejmg.2014.12.002.
23. Ekanem N, Ekure, Fidelia Bode-Thomas, Wilson E, Sadoh, Adeola A, Orogade et al. Congenital Heart Defects in Nigerian Children: Preliminary Data from the National Pediatric Cardiac Registry. *World Journal for Pediatric and Congenital Heart Surgery* 2017; Vol. 8(6) 699-706
24. Sadoh WE UC, Danies Q. Congenital heart diseases in Nigerian Children: a multicenter echocardiography study. *World Journal of Pediatric Heart Surgery* 2013;4(2):172-6.
25. Hwang IC, Sisavanh M, Billamay S, et al. Congenital heart disease at Laos Children's Hospital: two year experience. *Pediatr Int.* 2017;59(3):271-279.
26. Ratanasit N, Karaketklang K, Jakrapanichakul D, et al. Prevalence and echocardiographic characteristics of common congenital heart disease in adult patients at Siriraj Hospital: 10-year study. *J Med Assoc Thai.* 2015;98(1):7-13.
27. Bodian M, Ngaidé AA, Mbaye A, et al.: Prevalence of congenital heart diseases in Koranic schools (daara) in Dakar: a cross-sectional study based on clinical and echocardiographic screening in 2019 school children. *Bull Soc Pathol Exot.* 2015, 108:32-5.
28. Zhao QM, Liu F, Wu L, Ma XJ, Niu C, Huang GY: Prevalence of congenital heart disease at live birth in China . *J Pediatr.* 2019, 204:53-8. 10.1016/j.jpeds.2018.08.040.