



Original Research

Delays in Diagnosing Congenital Heart Disease: An Analysis of Contributing Factors in Patients Presenting to a Tertiary Care Hospital in Abbottabad, Pakistan

Article History:

Name of Author:

Zoya Fayaz¹, Saima Bibi², Ubaid Ali³,
Zainab Akhlaq⁴, Manahil Shamraiz⁵,
Rani Urooj⁶

Affiliation:

^{1,3,4,5,6}Post Graduate Resident, Department of
Paediatrics, Ayub Teaching Hospital,
Abbottabad, Pakistan.

²Associate Professor, Department of
Paediatrics, Ayub Teaching Hospital,
Abbottabad, Pakistan.

Corresponding Author:

Zoya Fayaz
Email: zoyafayaz1@gmail.com

Received: 04-11-2025

Revised: 14-12-2025

Accepted: 19-12-2025

Published: 30-12-2025

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-Noncommercial-Share Alike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

Abstract:

Objective: To determine the frequency of factors leading to delayed diagnosis of congenital heart disease in paediatric patients presenting to a tertiary care hospital. **Study Design:** Cross-sectional study. **Place and Duration of Study:** Conducted from 26 June 2025 to 26 October 2025 at the Department of Paediatrics, Ayub Teaching Hospital Abbottabad, Pakistan. **Methodology:** A total of 166 paediatric patients with delayed diagnosis of congenital heart disease were included in the study. Data regarding doctor-related, financial, referral, follow-up, social and midwifery related factors were obtained from parents, hospital records and previous prescriptions. Quantitative variables were expressed as mean $\pm$ standard deviation while qualitative variables were presented as frequency and percentage. Chi-square test and Fisher exact test were applied after stratification. **Results:** Mean age of patients was 6.41 ± 2.76 years, while mean age at diagnosis was 3.54 ± 2.36 years. Male patients were 94 (56.6%) and rural residents were 108 (65.1%). Acyanotic congenital heart disease was present in 110 (66.3%) patients. Financial factors were the most common contributor towards delayed diagnosis, found in 79 (47.6%) patients. Delayed hospital visit due to financial problems was noted in 58 (34.9%) cases. Referral and follow-up related factors were present in 61 (36.7%) patients, while delayed diagnosis by doctors was observed in 59 (35.5%) patients. **Conclusion:** Delayed diagnosis of congenital heart disease were common and mainly associated with financial difficulties, delayed referrals, rural residence and missed diagnosis by doctors.

Keywords: Congenital heart defects, Delayed diagnosis, Echocardiography, Heart diseases, Paediatrics.

INTRODUCTION

Congenital heart diseases represent one of the most prevalent types of congenital diseases among children, and can cause significant morbidity and mortality when not diagnosed early.¹ They involve congenital malformations involving structure of heart and great blood vessels. Symptoms associated with this type of disease include cyanosis, feeding problems, inadequate growth, rapid breathing, repeated infections of the chest or heart failure.² In some cases, symptoms can be very mild initially, with the disease going unnoticed for many years. Early diagnosis and management significantly improves outcome among patients suffering from this condition; nonetheless, delayed diagnosis remains

common among developing nations.³

The diagnosis of congenital heart defects is primarily based on thorough clinical assessment, proper referrals, echocardiography, chest X-rays, and electrocardiograms.⁴ In many instances, it becomes challenging to make the diagnosis because early signs and symptoms may not be specific and might be attributed to other diseases such as respiratory infection and malnutrition.⁵ Misdiagnosis by health professionals plays a vital role in delaying the detection of the condition. Patients may be misdiagnosed with pneumonia, asthma, or sepsis several times until the cardiac issue is detected. Lack of expertise among healthcare personnel and limited

access to pediatric cardiology services may delay the process of diagnosis.⁶

Social as well as economic variables have a very significant part to play in this regard. Individuals with low socio-economic status may experience a delay in seeking treatment at hospitals because of their financial difficulties, high medical bills, as well as high transportation charges.⁷ In remote locations, where there is less availability of specialized healthcare services, the travelling distance itself becomes one of the hurdles towards the early identification of the problem.⁸ Errors made while recording and understanding previous tests results, like that of an echocardiogram and chest radiography, can also lead to this delay in diagnosis.⁹

The delayed diagnosis of congenital heart disease is still considered to be one of the significant public health concerns in developing nations, which result in serious complications, multiple visits to hospitals, and death of young individuals. Local information about the underlying reasons like misdiagnosis by physicians, financial problems, transport costs, and false documentation of prior tests is scarce. Through this research, we will be able to highlight the most prevalent reasons for the delayed diagnosis of CHD.

Objective

The objective of this study is to analyse the contributing factors responsible for delays in diagnosing congenital heart disease.

METHODOLOGY

This cross-sectional study was conducted from 26 June 2025 to 26 October 2025 at the Department of Paediatrics, Ayub Teaching Hospital Abbottabad. Ethical approval for the study was obtained from the hospital ethical review committee before start of data collection with ethical certificate number Ref.No.RC-EA-2024/212. A total of 166 patients were included in the study. Sample size was calculated by using OpenEpi software while keeping 12.3% frequency of delayed diagnosis due to financial constraints,¹⁰ with 95% confidence interval and 5% margin of error.

Inclusion criteria

Patients of both genders aged up to 15 years presenting for first time with echocardiography confirmed diagnosis of congenital heart disease were included. Patients having cyanotic congenital heart disease not diagnosed within 1 week after birth and patients having acyanotic congenital heart disease not diagnosed within 3 months after birth were enrolled in the study.

Exclusion criteria

Patients having major congenital anomalies, chromosomal abnormalities, acquired heart diseases

including rheumatic heart disease and post viral myocarditis were excluded from study. Patients whose parents or guardians refused interview were also excluded.

Congenital heart defect was considered as any structural defect of heart present since birth causing signs and symptoms including cyanosis, tachypnoea, poor feeding, sweating during feeding, inadequate weight gain, recurrent respiratory tract infections, finger clubbing and cardiac murmurs confirmed on echocardiography. Cyanotic congenital heart disease was defined as congenital heart defect having right-to-left shunt with oxygen saturation less than 85% on pulse oximetry associated with clinical cyanosis. Acyanotic congenital heart disease was labelled as congenital heart defect with left-to-right shunt having oxygen saturation 85% or above without clinical cyanosis.

After taking informed consent from parents or guardians, confidentiality of patients was ensured and all information was recorded on specially designed proforma. Demographic details including age, gender, weight, socioeconomic status of family, educational status of parents and residential status were noted. Detailed history was taken from parents or guardians regarding factors causing delay in diagnosis of congenital heart disease. Patients were also assessed clinically and diagnosis of congenital heart disease was confirmed through echocardiography findings. Delay in diagnosis by doctors due to failure or delayed identification of the disease in previous health visits based on past prescriptions and previous records of hospitalization was taken into consideration. Delay in diagnosis by midwifery practitioners due to non-recognition of signs and symptoms of the disease in pregnancy, delivery, and puerperium was termed as such. The delay in seeking medical attention due to financial constraints or costs incurred from transportation expenses and the lack of money was categorized under financial barriers based on structured questionnaires. Delay in referring and following up patients after their initial visit was another barrier that can affect patient outcomes. Finally, social determinants consisted of education, residence, and birthplace of the parents, whether in government or private health care facilities.

Data was analysed by using IBM SPSS version 26. Quantitative variables including age were presented as mean $\pm$ standard deviation. Qualitative variables including gender, socioeconomic status, educational level of parents, residential status and factors contributing to delayed diagnosis were expressed as frequencies and percentages. Factors contributing to delayed diagnosis were stratified with respect to age, gender, socioeconomic status, educational level of

parents and residential status. Post-stratification chi-square test or Fisher exact test was applied where appropriate. A p-value of ≤ 0.05 was taken as statistically significant.

RESULTS

The study enrolled a total of 166 paediatric patients with congenital heart disease (CHD). The mean age of the patients were 6.41 ± 2.76 years, and the mean weight were recorded as 18.07 ± 7.15 kg. The average age at which diagnosis were made was 3.54 ± 2.36 years, which suggesting a notable delay between birth and formal identification of the condition. With regards to gender distribution, majority of the patients were male, accounting for 94 cases (56.6%), whereas female patients were 72 in number (43.4%). With respect to residential status, a considerable majority of the patients were from rural areas, accounting for 108 patients (65.1%), in contrast to urban residents who were 58 in number (34.9%). Regarding the type of CHD, acyanotic CHD were more prevalent, being present in 110 patients (66.3%), whereas cyanotic CHD were identified in 56 patients (33.7%) (Table 1).

Table 1. Patient Demographics

Demographics	n (%) or Mean $\pm$ SD
Age (Years)	6.41 ± 2.76
Weight (kg)	18.07 ± 7.15
Age at Diagnosis (Years)	3.54 ± 2.36
Gender	
Male n (%)	94 (56.6%)
Female n (%)	72 (43.4%)
Family Socioeconomic Status	
Low n (%)	82 (49.4%)
Middle n (%)	65 (39.2%)
High n (%)	19 (11.4%)
Educational Level of Parents	
Illiterate n (%)	37 (22.3%)
Primary n (%)	41 (24.7%)
Secondary n (%)	45 (27.1%)
Higher Secondary n (%)	35 (21.1%)
Graduate or Above n (%)	8 (4.8%)
Residential Status	
Urban n (%)	58 (34.9%)
Rural n (%)	108 (65.1%)
Type of CHD	
Cyanotic CHD n (%)	56 (33.7%)
Acyanotic CHD n (%)	110 (66.3%)

With reference to the factors which were contributing towards delayed diagnosis of CHD, financial factors were found to be the most frequently reported category, being present in 79 cases (47.6%). Among the financial sub-factors, delayed hospital visit due to financial reasons were noted in 58 cases

(34.9%), high medical costs were reported in 47 cases (28.3%), and transportation expenses were identified in 28 cases (16.9%). Referral and follow-up related factors were present in 61 cases (36.7%), with delayed referral to specialised care being the most common sub-factor at 47 cases (28.3%), followed by missed follow-up appointments in 30 cases (18.1%), and wrong reporting of previous investigations in 15 cases (9.0%). Doctor-related factors were also amongst the notable contributors, wherein delayed diagnosis by doctor were recorded in 59 cases (35.5%), and wrong or missed diagnosis by doctor were noted in 44 cases (26.5%). Social factors were present in 36 cases (21.7%), and delayed diagnosis related to midwifery care were reported in 27 cases (16.3%) (Table 2).

Table 2. Frequency of Factors Contributing to Delayed Diagnosis of Congenital Heart Disease

Factors	Frequency	%age
Doctor-Related Factors		
Delayed Diagnosis by Doctor	59	35.50 %
Wrong/Missed Diagnosis by Doctor	44	26.50 %
Financial Factors		
Financial Factors	79	47.60 %
Delayed Hospital Visit Due to Financial Reasons	58	34.90 %
High Medical Costs	47	28.30 %
Transportation Expenses	28	16.90 %
Referral & Follow-up Factors		
Referral/Follow-up Factors	61	36.70 %
Delayed Referral to Specialised Care	47	28.30 %
Missed Follow-up Appointments	30	18.10 %
Wrong Reporting of Previous Investigations	15	9.00 %
Other Factors		
Social Factors	36	21.70 %
Delayed Diagnosis Related to Midwifery Care	27	16.30 %

DISCUSSION

This research was designed for determining the causes behind the delay in the diagnosis of congenital heart diseases among children. The results of this research indicated that there is a very serious delay in making the correct diagnosis because the average age of diagnosis was found to be 3.54 ± 2.36 years, which is very late when compared to the optimal time

frame of early childhood. Financial factors appeared to be the most dominant cause of such delays, as they were found to be associated with 79 patients (47.6%). This result was expected because most of the patients belonged to lower socio-economic backgrounds, which comprised 82 patients (49.4%). The mean age at diagnosis in the present study were 3.54 ± 2.36 years, which is considerably higher than what were reported by Tahiri et al. 11 where mean age at diagnosis were only 3.28 days in neonatal population, and also higher than Kumari et al. 12 who reported mean age of 2.33 ± 3.81 years. This difference was likely because the present study included older paediatric patients rather than neonates, and also because majority of the patients were from rural backgrounds (108 cases, 65.1%) with limited access to early screening facilities, which is a barrier that were similarly highlighted by Al-Shawki et al. 13 who noted that diagnostic delays ranged from 4 to 98 months in resource-limited settings.

Acyanotic CHD were the predominant type in present study, being found in 110 patients (66.3%), which is consistent with findings of Tahiri et al. 11 who reported non-cyanotic CHD in 71% of their cases, and with Iqbal et al. 14 where acyanotic lesions were accounting for 67.9%. Similarly, Memon et al. 15 also reported VSD and ASD as the most common lesions. This similarity across different studies were suggesting that acyanotic lesions are inherently more difficult to detect early because they usually lack the obvious clinical features like cyanosis, and their subtle presentations such as murmurs or mild respiratory symptoms may easily get overlooked, particularly in settings where paediatric cardiology expertise is limited.

Financial factors were identified as the most common contributor to delayed diagnosis in the present study, being present in 79 cases (47.6%), which is in agreement with Shah et al. 16 who also reported financial limitations as a significant barrier, and with Al-Shawki et al. 13 who described socioeconomic limitations as a major obstacle in low- and middle-income countries. The high proportion of patients from low socioeconomic backgrounds (82 cases, 49.4%) in the present study were directly correlating with this finding, as families with limited financial resources often delays hospital visits due to inability to bear the cost of investigations and specialist consultations, a pattern which were also observed by Kumari et al. 12 and Iqbal et al. 14 in similar Pakistani tertiary care settings.

Rural residency was noted in 108 cases (65.1%) of the present study, and this finding were comparable to Kumari et al. 12 who reported rural residence in 63.6% of their patients, and to Rochmawati et al. 17

who found that remote residence was independently predicting delayed diagnosis with odds ratio of 1.82. Ng'eno-Owino 18 similarly reported that children from unskilled parental occupations, who were more likely residing in rural areas, had five times higher odds of late diagnosis.

Doctor-related factors were found in the present study, with delayed diagnosis by doctors reported in 59 cases (35.5%) and wrong or missed diagnosis in 44 cases (26.5%). These figures were closely comparable to Iqbal et al. 14 where missed or delayed diagnosis by doctors were present in 24.6%, and to Kumari et al. 12 who reported missed diagnosis by paediatricians in 34.1% of cases. This pattern of physician-level diagnostic delays was largely attributable to the limited availability of trained paediatric cardiologists, inadequate clinical examination skills at primary care level, and overreliance on symptomatic management without appropriate cardiac investigations, which is a concern that were also raised by Dewi et al. 19 and Chaka et al. 20 in their respective studies.

Low parental educational level was a notable finding in the present study, with illiterate and primary level educated parents together accounting for 78 cases (47.0%). This were consistent with Hurisa et al. 21 who found that illiterate maternal educational status were significantly associated with delayed diagnosis with AOR of 0.103, and with Ng'eno-Owino 18 who reported that parental unskilled occupation significantly increased odds of late diagnosis. Khan et al. 22 also highlighted that lack of knowledge regarding treatment options were present in 40.1% of parents. Low educational level amongst parents is strongly associated with poor recognition of cardiac symptoms, delayed health-seeking behaviour, and inability to follow up appropriately with specialist services, all of which were contributing collectively to the overall diagnostic delay observed in the present study.

There are various limitations of the current research that should be mentioned. First of all, this was a single-center study carried out at a single hospital which can be considered a serious limitation due to the fact that the obtained results cannot be extrapolated to the general population of Pakistan. Secondly, the sample size of 166 patients was relatively small which could affect the validity of the results. Finally, it was an observational study which prevents from establishing causality between the studied variables.

CONCLUSION

This study has shown that the late diagnosis of congenital heart diseases in children is a serious and multidimensional issue. Financial problems, rural residence, and lower education level of parents are considered the major causes for this problem. Factors related to the physician include delayed and missed diagnosis of congenital heart disease by doctors.

Disclaimer

None.

Acknowledgement

The authors are thankful to all doctors and departmental workers who gave support during study period. Their cooperation in maintaining records and managing patient information made this research possible to complete.

Ethical Approval

Approval for conducting this research was obtained from concerned ethical review committee. Whole research process was carried out according to committee instructions and Helsinki declaration principles.

Patient Consent

Written informed consent was obtained from every participant before inclusion in study. All patients were told that their personal information will be kept secret and they can leave the study whenever they want.

Conflict of Interest

The author stated that they have no conflict regarding this research work.

REFERENCES

1. Xu J, Li Q, Deng L, Xiong J, Cheng Z, Ye C. Global, regional, and national epidemiology of congenital heart disease in children from 1990 to 2021. *Front Cardiovasc Med*. 12:1522644. 2025. DOI: <https://doi.org/10.3389/fcvm.2025.1522644>
2. Lee J. Growth failure in children with congenital heart disease. *Children (Basel)*. 12(5):616. 2025. DOI: <https://doi.org/10.3390/children12050616>
3. Bai K, Wang T, Zhang S, He L, Wang Y, Ming X, et al. First-time detection of treatable congenital heart disease - a study of school-aged children in the community of three regions in China. *BMC Public Health*. 26:993. 2026. DOI: <https://doi.org/10.1186/s12889-026-26638-4>
4. Bakhshaliyev S, Karatza AA, Fouzas S, Gkentzi E. Diagnostic performance of cardiac CT and transthoracic echocardiography in congenital heart disease: a surgical correlation study. *Diagnostics (Basel)*. 16(2):259. 2026. DOI: <https://doi.org/10.3390/diagnostics16020259>
5. Karatza AA, Fouzas S, Gkentzi E, Varvarigou A. Missed or delayed diagnosis of heart disease by the general pediatrician. *Children (Basel)*. 12(3):366. 2025. DOI: <https://doi.org/10.3390/children12030366>
6. Ames S, Pillsworth E, Sparman-Shelto A, Isaac DL. Addressing barriers to health care access of congenital heart disease patients in Guyana. *Glob Pediatr Health*. 8:2333794X211012977. 2021. DOI: <https://doi.org/10.1177/2333794X211012977>
7. Greenwell AA, Deng MX, Ross S, Weixler V, Vervoort D. Socioeconomic status and access to care for pediatric and adult congenital heart disease in universal health coverage models. *J Cardiovasc Dev Dis*. 11(8):250. 2024. DOI: <https://doi.org/10.3390/jcdd11080250>
8. Vervoort D, Ross S, Weixler V, Greenwell AA. Global access to comprehensive care for paediatric congenital heart disease. *Children (Basel)*. 10(12):1896. 2023. DOI: <https://doi.org/10.3390/children10121896>
9. Jumhour H, Al-Shorafa A, Al-Hajri A. Delayed diagnosis in outpatient care: a systematic review of documentation fragmentation as a hidden driver of diagnostic error. *Cureus*. 18(2):e102990. 2026. DOI: <https://doi.org/10.7759/cureus.102990>
10. Rashid U, Qureshi AU, Hyder SN, Sadiq M. Pattern of congenital heart disease in a developing country tertiary care center: factors associated with delayed diagnosis. *Ann Pediatr Cardiol*. 9(3):210-5. 2016.
11. Tahiri F, Kadiri L, Danaoui K, Ettoini K, El Mahedaoui A, Zouine M, et al. Congenital heart disease: diagnostic and therapeutic challenges in clinical practice. *J Paediatr Adv Res*. 5(1):1-15. 2026. DOI: <https://doi.org/10.46889/JPAR.2026.5105>
12. Kumari V, Khan MA, Chand R, Ahsan AK, Khokhar RA, Shaikh AS, et al. Factors linked with the delayed diagnosis of congenital heart diseases. A single centre experience from a tertiary care hospital of Pakistan. *Prof Med J*. 30(10):1288-1292. 2023. DOI: <https://doi.org/10.29309/TPMJ/2023.30.10.7593>
13. Al-Shawki YMAB. The global burden and diagnostic challenges of undiagnosed congenital heart disease in resource-limited settings: a comprehensive systematic review from 2000 to 2025 of diagnostic gaps and solutions. 2025. DOI: <https://doi.org/10.21203/rs.3.rs-7663534/v1>
14. Iqbal S, Saidullah S, Ahmed RI, Khan MAA, Ahmed N, Khan MF. Factors contributing to delayed diagnosis of congenital heart disease in paediatric population. *Pak J Med Health Sci*. 15(5):1488-1491. 2021. DOI: <https://doi.org/10.53350/pjmhs211551488>
15. Memon ZA, Baloch D, Fareed M, Jokhio SA, Soomro SA, Ayoub Z, et al. Frequency and pattern

- of congenital heart disease in term neonates presenting in respiratory distress at tertiary care hospital, Larkana. *Pak J Health Sci*. 6(7):246-250. 2025. DOI: <https://doi.org/10.54393/pjhs.v6i7.3362>
16. Shah A, Bhalodiya P, Patel P, Trivedi B, Tandon K, Bhende V. Factors leading to delay in surgical treatment of paediatric congenital heart diseases in rural Western India – a hospital-based study. *Niger Med J*. 66(4):1623-1630. 2025. DOI: <https://doi.org/10.71480/nmj.v66i4.1014>
 17. Rochmawati ID, Fadlan MR, Martini H. Diagnostic delay in congenital heart disease related pulmonary arterial hypertension: insights from the single tertiary hospital centre of pulmonary hypertension registry in East-Java Indonesia. *Eur Heart J*. 43(Suppl):ehab849.113. 2022.
 18. Ng'eno-Owino MC. Factors associated with late diagnosis of congenital heart disease in Kenya. Nairobi: Aga Khan University; 2018.
 19. Dewi PDR, Gunawijaya E, Yantie NPVK. Early versus late diagnosis of critical congenital heart disease at Sanglah Hospital Denpasar, Bali. *GSC Adv Res Rev*. 11(3):72-76. 2022. DOI: <https://doi.org/10.30574/gscarr.2022.11.3.0159>
 20. Chaka TE, Ali HA, Tadele H. Delayed diagnosis of congenital heart diseases and associated factors in the largest tertiary hospital in Ethiopia. *Cardiovasc Diagn Ther*. 15(5):955-965. 2025. DOI: <https://doi.org/10.21037/cdt-2025-186>
 21. Hurisa T, Megersa H, Tsegaye T. Delayed in diagnosis of congenital heart disease and associated factors among paediatric patients in Cardiac Centre Addis Ababa, Ethiopia, 2021/2022 G.C. *Am J Health Res*. 10(3):51-62. 2022. DOI: <https://doi.org/10.11648/j.ajhr.20221003.12>
 22. Khan MJ. Parental awareness and delays in seeking surgical care for paediatric congenital anomalies. *J Popul Ther Clin Pharmacol*. 18(3):e567-e574. 2011.