Original Research Article

Clinical suspicion of congenital heart diseases during well-baby follow-up and echocardiographic correlation

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ABSTRACT

Background: Congenital heart disease (CHD), the most common cardiac malformation in the fetal and neonatal period, represents a heterogeneous group of defects with little known cause. Despite advances in detection and treatment, CHD accounts for 3% of all infant deaths and 46% of death from congenital malformations. CHD patients with minimum or no symptoms are frequently under diagnosed in areas with inadequate health services. The aim of this study was to investigate the role of clinical suspicion of congenital heart disease in babies who presented for well-baby follow-up against echocardiographic findings and to study the clinical spectrum of all the CHD that were diagnosed.

Methods: The study involved analysis of data collected by the authors while working in a tertiary care center at Mumbai, India, during 2009 to 2010. All the babies born from April 2009 to August 2010 (17 months) and those who came for well-baby visits (with or without symptoms) were examined carefully for findings on cardiac examination including abnormalities of heart sound and presence of murmur on auscultation. Findings of the echocardiography were recorded for follow up and all the babies were followed up on at least three-monthly basis. Echocardiography was repeated on a quarterly basis in symptomatic babies and six-monthly basis in asymptomatic babies with small septal defects. All the babies on follow up were closely monitored clinically and on echocardiography for ascertaining the natural course of the observed defects.

Results: A total of 1856 babies were born at the tertiary care center out of which 1688 were seen at the well-baby clinic. A total of 42 cases were referred for echocardiography on clinical suspicion of CHD and out of these 21 (50%) were positive for some form of CHD. Most common CHD diagnosed was atrial septal defect (ASD). There was no spontaneous closure of ventricular septal defects (VSD), though, 02 cases of ASD less than 3 mm closed during the follow up period.

Conclusions: This study demonstrated that clinical experience is a valuable tool in early diagnosis of CHD, even if they are asymptomatic. Early echocardiography is justified to diagnose the cases so that appropriate follow-up and management decisions could be planned. Though majority of the cases were moderate to small ASD and VSD, there is a need to diagnose them early and follow them up till closure.

Keywords: Congenital heart disease, Echocardiography, Well baby visit

INTRODUCTION

Congenital heart disease (CHD), the most common cardiac malformation in the fetal and neonatal period, represents a heterogeneous group of defects with little

known cause. Congenital heart defects are the most common of all congenital malformations, with an incidence at 6 to 8 per 1000 live births. The Centers for Disease Control reports cyanotic heart defects occurred in 56.9 per 100,000 live births in the United States in 2005,

with higher rates noted when maternal age exceeded 40 years. The incidence of congenital heart disease in premature infants is 12.5 per 1000 live births, excluding isolated patent ductus arteriosus (PDA) and atrial septal defect (ASD). Despite advances in detection and treatment, CHD accounts for 3% of all infant deaths and 46% of death from congenital malformations.¹ CHD patients with minimum or no symptoms are frequently under diagnosed in areas with inadequate health services.² The aim of this study was to investigate the role of clinical suspicion of congenital heart disease in babies who present for well-baby follow-up against echocardiographic findings and to study the clinical spectrum of all the CHD that were diagnosed. It was also designed to study the natural course of the lesions detected during the follow up period on a continuum.

METHODS

The study involved analysis of data collected by the authors while working in a tertiary care center at Mumbai, India, during 2009 to 2010. All the babies born from April 2009 to August 2010 (17 months) and those who came for well-baby visits (with or without symptoms) were examined carefully for findings on cardiac examination including abnormalities of heart sound and presence of murmur on auscultation. Based on the Nadas criteria and clinical experience of the primary author, only the babies in whom the cardiac findings and murmurs that appeared unlikely to be of functional origin were referred for echocardiography (with Doppler study wherever applicable) at Cardiology OPD. Two days in a week were reserved for routine Pediatric echocardiography by the cardiologists for speedy evaluation with minimum delay in diagnosis.

Sedation of the babies for echocardiography was carried out by administering 50-75 mg/kg of Triclofos under monitoring. Mothers were interviewed for any symptoms in the baby that could suggest cardiac disease like failure to thrive, increased rate of breathing, feeding difficulty and excessive sweating. Findings of the echocardiography were recorded for follow up and all the babies were followed up on at least three-monthly basis. Echocardiography was repeated on a quarterly basis in symptomatic babies and six-monthly basis in asymptomatic babies with small septal defects. All the babies on follow up were closely monitored clinically and on echocardiography for ascertaining the natural course of the observed defects. The results were tabulated and descriptive analysis of the findings carried out.

RESULTS

During the study period from April 2009 to August 2010, a total of 1856 babies were born at the tertiary care center out of which 1688 were seen at the well-baby clinic. A total of 42 cases were referred for echocardiography on clinical suspicion of CHD and out of these 21 (50%) were positive for some form of CHD (Figure 3), though few of

them were only small septal defects. The study excluded neonates diagnosed with small patent foramen ovale. Five babies with CHD were preterm, with birth weight between 2-2.4 kilograms. This gives the prevalence of CHD seen in this cohort of babies as 12.4% (21/1688). The age and sex distribution of the CHD cases is shown in Table 1. Various types of CHD diagnosed in these babies is highlighted in Figure 1.

Table 1: Age and sex distribution of babies with CHD.

Characteristics	Number
Total No of babies referred for Echocardiography	42
Total number of babies diagnosed with CHD on Echo	21
Male: Female ratio	13:8
Age group of babies with CHD	
<1 month	07
1-3 month	07
3-6 month	04
>6 months	03

Details of symptomatic cases and asymptomatic cases picked by careful clinical examination are given in the Table 2. Two cases with ASD size more than 8mm, one baby with a cyanotic CHD and one child with ASD and a sub-aortic VSD (6.7mm) became symptomatic around three months of age with feeding difficulty and slow weight gain. They were treated with anti-failure medication with digoxin and furosemide and they thrived well on medication.

Figure1: Types of CHD seen in the babies.

ASD: Atrial Septal Defect (ASD <3mm: 03, 3-8 mm: 06, >8mm :02); VSD: Ventricular septal defect; PDA: Patent ductus arteriosus; Diagnosis of causes under category 'others' included: Valvular Pulmonary Stenosis (01), Bicuspid aortic valve with post-ductal coarctation (01), Single ventricle with Pulm Stenosis and PDA (01), Double outlet right ventricle with inlet VSD and PS (01).

All the babies were regularly followed up clinically and 3-6 monthly echocardiographic evaluation till the end of the study period. Follow-up details with natural course of the detected lesions are given in Table 3.

Table 2. Symptomatic versus asymptomatic babies during study period.

Characteristics	No.
Completely asymptomatic at diagnosis	20
Mild nail bed and mucosal cyanosis with tachypnoea on day 1	01
Became/remained symptomatic during follow up by three months	04
Total babies who remained asymptomatic till July 2010	17

Table 3: Natural course of the detected CHD lesions during study period.

Characteristics	No.
Total number of children with CHD	21
Total lost to follow up by end of the study period (Transfer, return to hometown)	05
Spontaneous ASD closure by 03 months	Nil
Spontaneous decrease in ASD size (01 case<3mm)	02
Spontaneous closure of ASD with no shunt (both <3mm)	02
Spontaneous PDA closure by 03 months	01
Spontaneous decrease in VSD size	01
Spontaneous closure of VSD	Nil

DISCUSSION

It is not infrequent that one comes across diagnosing and treating small infants with CHD. These defects pose considerable challenge in counseling the parents about prognosis and need meticulous follow up and intervention at appropriate time to prevent mortality and morbidity. Hearing murmur in children during routine examination is a common event. Most often, when the child is asymptomatic and murmur is less than grade III one tends to dispose it off as functional. With routine auscultation regularly and with experience, it is possible to identify murmurs with significance. This is aptly evident in the present study as 50% (21/42) of the babies referred for echocardiography were detected to have one of the CHD.

The overall prevalence of CHD based on most of the available literature is 8/1000 liveborn. A total of 21 cases diagnosed in 1688 deliveries makes a prevalence rate of 12.4/1000 in this series. This increase could possibly be due to early diagnosis of small ASD, even less than 3mm in size. In a large study in Kanpur, a prevalence rate of 26.3/1000 of CHD was observed.³ Saleh et al, reported a rate of over 39% CHD when symptomatic children were referred for echocardiography.⁴ Capozzi et al also reported a regional incidence rate of 11.1 per thousand livebirths on one of the largest series studied involving over 4 lakh liveborns.² In this regard, 50% detection rate of CHD among predominantly asymptomatic babies referred for echocardiography in the present study strongly favors the role of clinical approach in early detection of congenital heart diseases.

There was a male preponderance in the present series (13 versus 8). It is difficult to explain the reason for this preponderance with the available evidence. Such male preponderance has also been observed by other workers.^{5,6} Eighty six percent (18/21) of the cases in the present series were diagnosed within six months and majority of them were asymptomatic. This highlights the importance of strong clinical suspicion for CHD and role of early echocardiography in identifying these cases. As echocardiography is a non-invasive process and needs only moderate sedation in small babies, every baby with strong clinical suspicion of CHD should undergo an echocardiography, even if asymptomatic at presentation. Bostan et al diagnosed ASD and other interatrial communications in 847 newborns by involving large number of babies in the screening and the mean age at diagnosis was an astonishing 1.7 days (range: 1-7 days).⁷ In another study, the median age at diagnosis of ASD was 5 months with a range of day-1 to 14 years.⁸ Quite often diagnosis of ASD is missed till adulthood or it is incidentally detected, hence, early echocardiography on clinical suspicion should be strongly advocated to pick these cases.

The most common diagnosis in the present series was ASD (11/21) followed by VSD (3/21). Two cases had both ASD and VSD. One child had small multiple VSDs. Majority of the cases (19/21) were acyanotic CHD. In a series by Akhtar et al, 19% of the cases were cyanotic CHD.⁵ In their series, VSD was the most common diagnosis followed by PDA and ASD. Few other recent studies also have seen VSD as the most common acyanotic CHD.⁹ ASD as the most common diagnosis in the present study is likely due to early screening of asymptomatic babies with cardiac findings for CHD, with more cases of moderate to small ASD diagnosed in the present series. ASD as the most common diagnosis (53%) was also reported by Hannoush et al.¹⁰ Both cyanotic patients were symptomatic at diagnosis. Two patients, one with a large VSD and other with an ASD along with sub-aortic VSD needed treatment with decongestive measures by four months of age due to progressive symptoms. The cases were well controlled with medical therapy alone.

Rate of spontaneous closure or spontaneous reduction in size of the septal defects in this series was 36% (4/11) for ASD and 0% (0/3) for VSD. Size of one VSD reduced with decreased shunt during follow up, none of them closed spontaneously. One case of PDA also spontaneously closed by 03 months of age. Two cases of ASD with diameter less than 3 mm closed by 06 months and one ASD <3 mm reduced in size to only shunt detectable on color Doppler during follow up. None of the ASD >3mm closed during the follow up period, though, most of them remained asymptomatic and thrived well. These findings are consistent with the observation that small ASD with dimension <3-4 mm is likely to close spontaneously and do not need follow up.⁷⁻¹¹ It is recommended that all interatrial communications that are

more than 3 mm, even if asymptomatic are to be followed up till complete closure.⁷

Aptly a question arises as to what is the relevance of this study if majority were asymptomatic ASD. Cryptogenic ischemic events occur in young patients and have serious sequelae. The potential for paradoxical embolization through a PFO or an ASD should be assessed in all such patients.¹² Bannan et al in a study published during 2009 demonstrated that 20 patients with ASD (14%) presented with a paradoxical embolism. These patients tended to be younger and had smaller defects (both by size and shunt ratio) than ASD patients without paradoxical embolism, and were more likely to be female than PFO patients. They concluded that incidence of adult patients with ASD who presented with paradoxical embolism is higher than expected and suggests that this diagnosis should be considered in patients with cryptogenic stroke.¹³ Giardini et al demonstrated that in patients with PFO, migraine headache with aura is associated with spontaneous large R-to-L shunt and thrombophilic conditions. Transcatheter defect closure seems to be an effective and safe means to treat migraine headache with aura in patients with PFO.¹⁴ Early diagnosis of these lesions also helps the clinician to look for other associations. In view of these observations, it is clear that diagnosis and follow up of CHD with even small defects is of paramount importance in the long run. Moreover, it is felt that any tertiary care center with facilities to carry out echocardiography and doppler should make all out efforts to diagnose cases presenting to their facility, however small the defect may be.

CONCLUSION

In conclusion, this study demonstrated that clinical experience is a valuable tool in early diagnosis of CHD, even if they are asymptomatic. Early echocardiography is justified to diagnose the cases so that appropriate follow-up and management decisions could be planned. Though majority of the cases were moderate to small ASD and VSD, there is a need to diagnose them early and follow them up till closure. Early diagnosis also helps in identification of other associations and to treat the cases before the symptoms reach an advanced stage.

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