

The Pattern of Congenital Heart Disease among Neonates Referred for Echocardiography

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ABSTRACT

Objective: To define the pattern of congenital heart disease (CHD) among neonates referred for echocardiography.

Setting: Department of Pediatrics, Qatif Central Hospital.

Design: A Prospective longitudinal and hospital based study.

Method: All neonates with suspected CHD referred for echocardiography were reviewed for one year from March 2011.

Result: Echocardiography was requested for 289 (9.5%) neonates, male to female ratio 1:1.1, mean birth weight 2.909 kg (range 0.605-5.150 kg); most of them were full-term (86%). One hundred forty-six (50.5%) neonates underwent echocardiography within the first 24 hours (group I).

Among those neonates referred for echocardiography, 245 (84.8%) had heart murmur, 9 (3.1%) had cyanosis, 5 (1.7%) had questionable cardiomegaly, 13 (4.5%) had congenital anomalies, and 17 (5.9%) had various anomalies. Heart murmur was detected in 257 (88.9%) during routine neonatal examination.

Sixty-four (2.1%) neonates were diagnosed to have CHD, 8 (12.5%) of them have critical congenital heart disease (CCHD). The most common CHD was ventricular septal defect (VSD) 32 (43.2%) followed by secundum atrial septal defect 31 (41.9%). Among 23 neonates with congenital anomalies referred for echocardiography, 10 (43.5%) had Down syndrome. Some neonates had more than one anomaly.

Conclusion: Most neonates suspected to have CHD were referred for echocardiography during the first 24 hours. Heart murmur was the most common indication of referral for echocardiography and most of the cases with CHD and CCHD presented with heart murmur alone. Almost all cases of CCHD were diagnosed within the first 72 hours. The most common CHD was VSD followed by secundum ASD. Down syndrome was the most common congenital anomaly referred for echocardiography and 30% of them have CHD.

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INTRODUCTION

Congenital heart disease (CHD) occurs in 0.8% of live births. The incidence is higher in premature infants, about 2% excluding patent ductus arteriosus (PDA). The diagnosis is established by one week of age in 40%-50% of patients with CHD. Despite the advances in palliative and corrective surgery, CHD remains the leading cause of death in children with congenital malformations. Most of CHD is ventricular septal defect (30%-35%); about one quarter could be critical congenital heart disease (CCHD) who would require cardiac surgery or catheterization before the age of one year^{1,2}.

As CHD associated with significant morbidity and mortality, determining the prevalence and pattern of CHD is necessary to recommend valuable changes in health policies including cost effective medical and surgical management and preventive measures.

The aim of this study is to determine the pattern of CHD among neonates referred for echocardiography.

METHOD

This longitudinal and hospital-based study was performed for live births for one year which started in March 2011.

Neonates delivered were examined by pediatric resident then confirmed by neonatologist if there was suspicion of CHD. The following information were recorded: postnatal age at time of echocardiography, gender, nationality, term/preterm delivery, birth weight, O₂ saturation, congenital anomalies if present, heart murmur, and suspected cardiomegaly from chest X-ray (CXR). Neonatal age group has been classified according to the time of echocardiography; group I within the first 24 hours of life, group II between 24-48 hours, group III older than 48 hours till 72 hours and group IV older than 72 hours.

Echocardiography has been performed randomly by one of two certified pediatric cardiologists in service using one of the following echocardiography machines: VIVID E7 or E9, Philip CX-60.

RESULT

A total of 3,033 babies were born during one year period (Saudi 95%, non-Saudi 5%). Echocardiography was requested for 289 (9.5%). Male to female ratio was 1:1.1, the mean birth weight was 2.909 kg, a range of 0.605-5.150 kg; 248 (86%) were full term, while preterm were 41 (14%), see table 1.

Table 1: Birth Characteristics

Demographic data	Number and Percentage
Total live born neonates	3,033
Nationality:	
• Saudi	2,877 (95%)
• Non Saudi	156 (5%)

Neonates referred for echo	289 (10%)
Gender:	
• Male	139 (48%)
• Female	150 (52%)
Mean birth weight (range)	2.909 (0.605-5.150) kg
Term/Preterm (ratio)	248/41 (6:1)

One hundred forty-six (50.5%) neonates had echocardiography within the first 24 hours (group I) and was considered the most common age group referred for echocardiography. The rest of neonates were in the following age group sequence: group II: 66 (22.8%), group III: 36 (12.5%), group IV: 41 (14.2%), see figure 1.

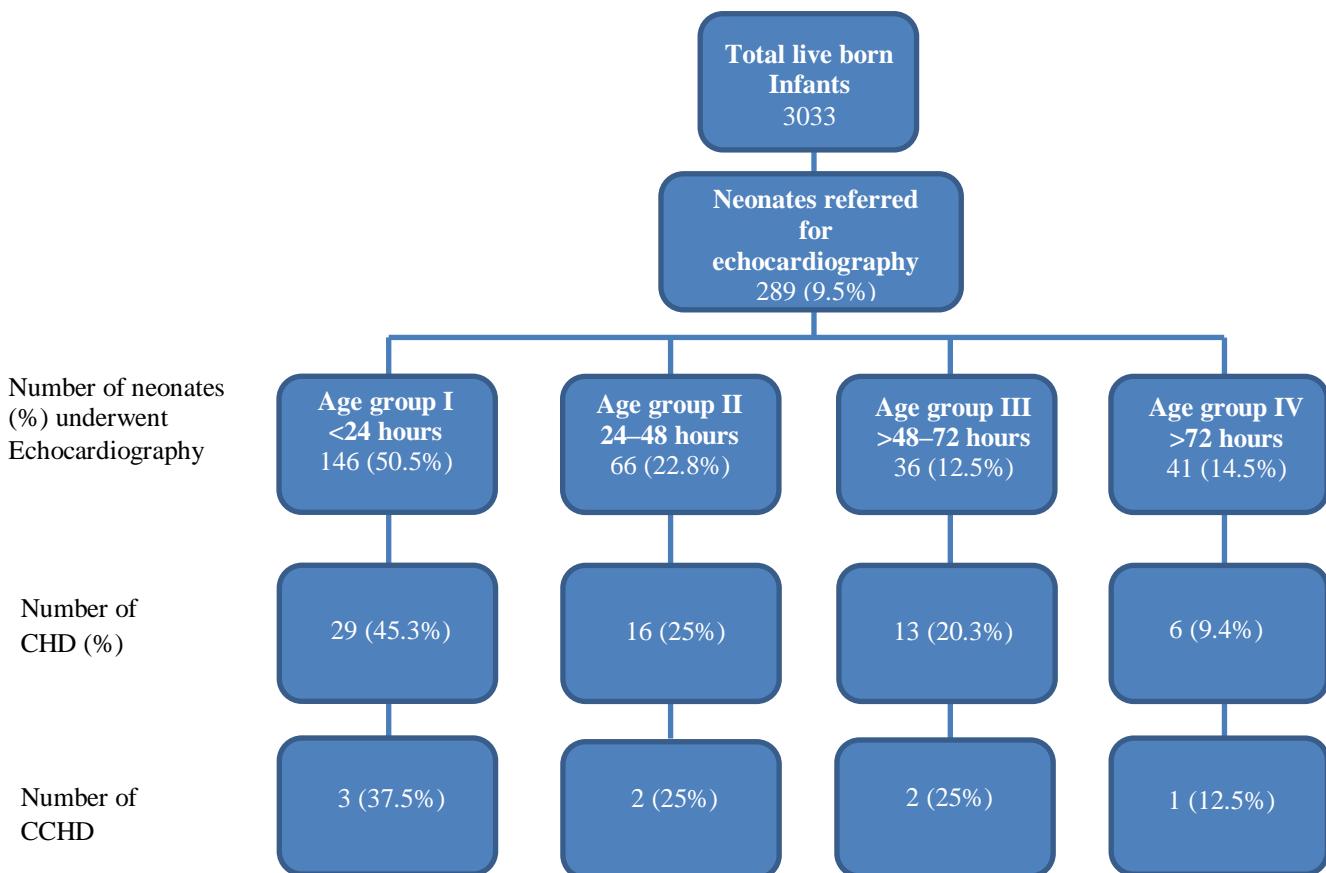


Figure 1: Neonatal Age Group and Frequency of Congenital Heart Disease (CHD) and Critical Congenital Heart Disease (CCHD)

Among those neonates referred for echocardiography, 245 (84.8%) neonates had heart murmur, 9 (3.1%) neonates had cyanosis, 5 (1.7%) neonates had questionable cardiomegaly, 13 (4.5%) neonates had congenital anomalies and 17 (5.9%) neonates had various anomalies. Overall, heart murmur (alone or combined with other indications) detected in 257 (88.9%) during routine neonatal screening was the most common indication for echocardiography.

After excluding PDA (193 neonates) which might reflect a transient physiologic pattern, 64 (22.1%) live born infants were diagnosed to have CHD; 52 (81.3%) of them were referred merely for heart murmur, see figure 2. The most common CHD was ventricular septal defect (VSD) (43.2%) followed by secundum atrial septal defect (ASD II), see table 2. Fifty-eight out

of sixty-four (90.7%) neonates were diagnosed to have CHD within the first 72 hours of life, see figure 2.

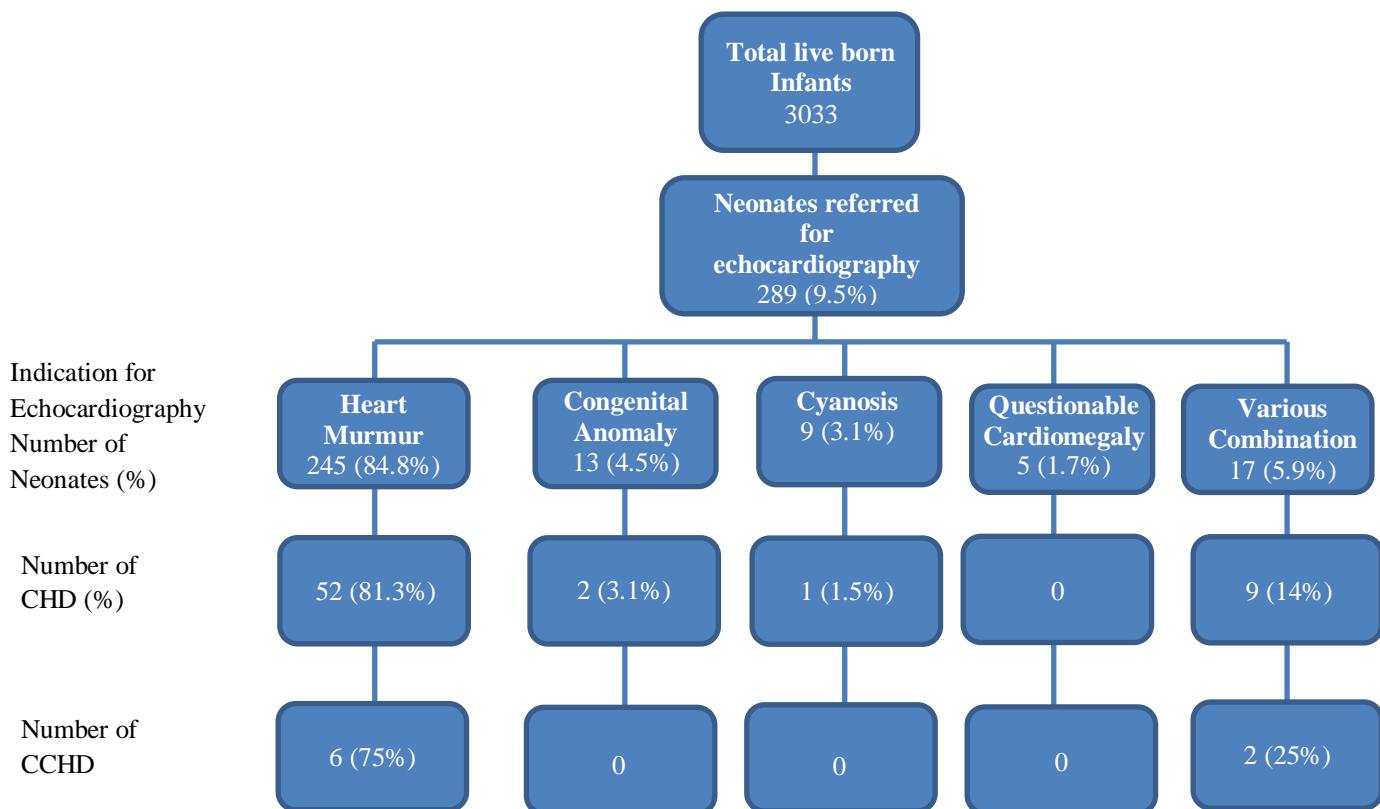


Figure 2: Indications of Referral for Echocardiography and Frequency of Congenital Heart Disease (CHD) and Critical Congenital Heart Disease (CCHD)

Table 2: Congenital Heart Disease (CHD), excluding Patent Ductus Arteriosus

CHD	Number and percentage
Ventricular septal defect	32 (43.2%)
Secundum atrial septal defect	31 (41.9%)
Pulmonary valve stenosis*	4 (5.4%)
Complete atrioventricular septal defect	2 (2.7%)
Other CHD**	5 (6.8%)

* One case of mild stenosis, 2 cases of severe stenosis, one case of critical stenosis.

** Dextrotransposition of great arteries, tetralogy of Fallot, coarctation of aorta, tricuspid atresia, hypoplastic left heart syndrome.

*** Some neonates had more than one anomaly.

CCHD was diagnosed in 8 neonates (12.5% of CHD) which include: dextro-transposition of great arteries, tetralogy of Fallot, critical pulmonary stenosis, hypoplastic left heart syndrome, tricuspid atresia, coarctation of aorta in addition to two cases of severe pulmonary stenosis. All CCHD cases except one were diagnosed within the first 72 hours of life (group I-III). Six cases of CCHD presented only with heart murmur while the other 2 cases presented with multiple symptoms and signs, see figure 2.

The total number of reported congenital anomalies referred for echocardiography was 23 cases, 10 (43.4%) of them had Down syndrome. After exclusion of PDA (which was found in all cases of Down syndrome and may reflect a transient physiologic pattern), 3 (30%) neonates

with Down syndrome were diagnosed to have CHD [two cases have VSD and one case has complete atrioventricular septal defect (AVSD)]. There were no CHD among neonates with other congenital anomalies (5 cases skeletal anomalies, 2 cases prune belly syndrome, 2 cases meningomyelocele, 2 cases cleft lip and palate, one case Apert syndrome and one case Leprechaunism syndrome).

DISCUSSION

Neonates with CHD may be diagnosed on the basis of physical examination. Normal examination does not exclude CHD and some studies showed that routine neonatal examination fails to detect more than half of the babies with CHD³. The accuracy of skilled physical examination to distinguish between pathological and innocent heart murmur is limited even for certified neonatologists and cardiologists; the prevalence of heart murmur in newborn infants is 0.6%- 4.2%³⁻⁶. Heart murmur is not always accompanying CHD; about half of the neonates with heart murmur have CHD⁷. In our study, heart murmur was detected in 8.5% of neonates, it was the most common indication of referral for echocardiography; 81.3% of neonates with heart murmur were found to have CHD.

The American Heart Association supports the Secretary of Health and Human Services' recommendation that pulse oximetry should be used to screen newborns for CCHD. Oximetry assessment performed after 24 hours of life showed that the sensitivity for detecting CCHD was 69.6%, and the positive predictive value was 47%⁸. In our study, 9 (3.1%) neonates were referred for the presence of cyanosis alone but only one case was found to have non-CCHD.

Radiologic assessments of the chest radiographs of children and infants having asymptomatic heart murmur revealed low reproducibility and low accuracy for detecting heart disease even if the interpretation was made by radiologists⁹⁻¹¹. In our study, 5 (1.7%) neonates with questionable cardiomegaly were referred for echocardiography and none of them have CHD.

Worldwide, about 40% of Down syndrome has CHD and 50% of the defects are AVSD¹². In our study, CHD was found in 30% of neonates with Down syndrome (VSD 20% and AVSD 10%).

Echocardiography has serious limitations as a screening tool, including its cost, high frequency of false positive results or recognition of clinically benign diagnosis¹³.

In our study, most neonates with CHD and CCHD were discovered within the first 72 hours and it is anticipated that significant cases could be missed with earlier neonatal discharge.

The most important limitation in our study is the number of the newborn population and this can be improved if the study performed over a longer period or performed as a multi-centric study. To have valuable insights for discovering possible missed cases during clinical evaluation at birth and proper prevalence of CHD, a follow-up clinical evaluation at age of 6 weeks is required³.

CONCLUSION

Most neonates suspected to have CHD were referred for echocardiography during the first 24 hours. Heart murmur was the most common indication of referral for echocardiography and most of the cases with CHD and CCHD presented with heart murmur alone. Almost all cases of CCHD diagnosed within the first 72 hours. The most

common CHD was VSD followed by secundum ASD. Down syndrome was the most common congenital anomaly referred for echocardiography and 30% of them have CHD.

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