Birth prevalence of congenital heart disease: A cross-sectional observational study from North India

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ABSTRACT

Objective : **To assess the birth prevalence and pattern of congenital heart disease (CHD) using echocardiography in babies born in a community hospital of North India.**

Methods : **A cross-sectional observational study conducted over a period of 3 years. Newborns born over a specific 8-h period of the day were recruited in the study. They underwent routine clinical examination and pulse oximetry, followed by screening echocardiography for diagnosing a CHD.**

Results : **A total of 20,307 newborns were screened, among which 874 had abnormal echocardiograms; 687 had insignificant CHDs, 164 had significant CHDs, and 24 had other abnormal cardiac findings. The birth prevalence of significant CHDs was 8.07 per 1000 live births; 131 newborns had an acyanotic CHD (79.9%) and 33 a cyanotic CHD (20.1%). Ventricular septal defect (VSD) was the most common acyanotic CHD, present in 116 newborns, giving a prevalence of 5.7/1000 live births. Among the cyanotic CHD, transposition of great arteries was most common (prevalence 0.34/1000 live births).**

Conclusion : **The CHD birth prevalence in our study is similar to the reported worldwide birth prevalence. Acyanotic CHD (mostly VSD) is seen in about three-fourths of babies born with CHD. The more sinister cyanotic CHD is present in remaining 25%.**

Keywords : Congenital heart disease, echocardiography, newborns, prevalence

INTRODUCTION

Congenital heart disease (CHD) has been defined as a gross structural abnormality of the heart or intrathoracic great vessels that is actually or potentially of functional significance.[1] In the developed world, CHD is considered to be the most major congenital anomaly and a leading cause of mortality in the first year of life.[2,3] However, little data is available from developing countries. The birth prevalence of CHD is estimated to be eight per 1000 live births.[4,5] The burden of CHD in India is likely to be enormous, because of a very high birth rate. It is estimated that over 180,000 children in India are born with CHD every year.[6] As only a very small proportion get required intervention, the number of young adults with CHD is steadily increasing. This heavy burden emphasizes the importance of CHD in India.

Recent advances in cardiovascular diagnostics and therapeutics have increased the survival of infants and

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children with CHDs. Unfortunately, access to quality care for children with CHD is largely restricted to developed parts of the world. It is important to have reliable information about CHD birth prevalence in the developing world, including India, to plan and provide better care to these patients. An earlier study from India,[7] published in 1994, has reported on the prevalence of CHD at birth, but the diagnosis is primarily based on clinical examination. It is well known that routine clinical examination of newborns has a poor sensitivity for detection of CHD.[8,9] Echocardiography with Doppler is the gold standard for the diagnosis of CHD in newborns with a very high sensitivity and specificity.[10-12]

In our study, we describe the birth prevalence of CHDs in babies born in a general hospital of North India using screening echocardiography.

METHODS

In a cross-sectional observational study conducted over a period of 3 years at the Safdarjung Hospital located in North India, we screened newborns for CHDs using echocardiography. Nearly 20,000 deliveries occur in this hospital every year, but babies born over a period of 8 h everyday were included in the study for logistic reasons. Newborns were screened for CHDs by a study team comprising of two pediatric cardiologists, a research officer, a field investigator, and a social worker. The team was from the All India Institute of Medical Sciences (AIIMS), a tertiary referral center located very close to the Safdarjung Hospital.

The ethics committees of both the primary hospital and the referral center approved the study protocol.

Informed consent

An informed consent sheet with details of the study protocol was given to one of the parents and approval sought in writing before recruiting the newborn in the study. None of the parents refused to provide consent.

Initial evaluation

The research officer performed routine clinical examination within 24 h of birth. This was recorded in a form that included the following parameters: central cyanosis, murmur on chest auscultation, and respiratory distress. Also the field investigator for all newborns obtained noninvasive arterial oxygen saturation within 48 h of life. Oximetry values were obtained from one of the feet of the baby. A persistent saturation of less than 95% was considered abnormal.

Echocardiography

All newborns recruited into the study were screened using transthoracic echocardiography after the initial evaluation, but within 48 h of life. A pediatric cardiologist performed echocardiography using the Fujifilm SonoSite M Turbo (Bothell, WA, USA) ultrasound system. The technique involved performing cross-sectional echocardiography, and Doppler and color flow imaging in various views. The cardiologist was not aware of the results of the initial clinical evaluation. All loops and images were stored in digital format for later review, if required. Newborns with CHDs were referred to AIIMS for further management or intervention, if needed. Also in case of any difference in diagnosis, the test was repeated at AIIMS by one of the pediatric cardiologists using the Philips IE-33 (Philips echocardiography equipment, Philips Inc., Bothell, WA, USA).

Definition of congenital heart diseases

1. **Normal:** No echocardiographic abnormality or any of the following: Patent ductus arteriosus (PDA) <2 mm in size without volume overload of left ventricle, patent foramen ovale or atrial septal defect (ASD) <5 mm without volume overload of right ventricle, and mild turbulence at branch pulmonary arteries.
2. **Insignificant congenital heart diseases:** Very small muscular ventricular septal defect (VSD), as these are likely to close spontaneously.
3. **Significant congenital heart diseases:** These are divided into minor and major CHDs:
   a. **Minor congenital heart disease:** ASD >5 mm, PDA >2 mm with left ventricle volume overload, restrictive VSD, and valvular aortic/pulmonary stenosis with gradients <25 mmHg.
   b. **Major congenital heart disease:** Further divided into acyanotic and cyanotic CHDs; any CHD that was likely to require intervention within the first year, including newborns with critical CHD that require intervention within the first 4 weeks of life.
      i. **Acyanotic CHD:** Nonrestrictive VSD, valvular aortic/pulmonary stenosis with gradients >25 mmHg, and coarctation of aorta (nonduct dependent).
      ii. **Cyanotic CHD:** Hypoplastic left heart syndrome, transposition complexes, aortic arch interruption, univentricular heart, Tetralogy of Fallot, Tetralogy of Fallot-like conditions associated with pulmonary stenosis or atresia, total anomalous pulmonary venous connection, persistent truncus arteriosus, Ebstein's anomaly, etc.
4. **Other cardiac findings on echocardiography:** Persistent pulmonary hypertension of newborn, cardiac tumor, situs inversus dextrocardia, and arrhythmias (complete heart block, supraventricular tachycardia).

Statistical analysis

The sample size was calculated by using the estimates for prevalence of CHD as 6-8/1000 live births. A sample size

of 6000 was calculated to be adequate. However, as one of the secondary objectives was to determine the relative proportion of various congenital heart defects, especially the major ones, the sample size chosen was 20,000. This is in keeping the incidence of critical heart disease as three per 1000 live births. A sample size of 20,000 will provide 160 newborns with congenital cardiac defects and about 60 of these will have major CHD requiring early intervention. Data was managed and entered in MS Excel and analyzed using Stata 11.0 (College Station, Texas, USA). The prevalence rate was reported as "per 1000 live births"; 95% confidence intervals (CIs) were calculated using the Wilson method.

RESULTS

A total of 20,307 newborns were recruited in our study and screened with echocardiography for diagnosing a CHD. Among these, 10,878 were boys and 9429 were girls. The birth weight ranged from 0.7 to 5.2 kg (mean 2.79 ± 0.53 kg). The echocardiographic findings are shown in the Figure 1.

Birth prevalence of congenital heart diseases

In the total cohort, 874 newborns had an abnormal echocardiogram at birth. Among these, 164 had significant CHDs, yielding a birth prevalence rate of 8.07 per 1000 live births (95% CI, 6.94-9.40 per 1000); 131 babies had acyanotic CHD, of which 93 had a minor CHD and 38 a major acyanotic CHD, yielding a birth prevalence of 4.58 (95% CI, 3.74-5.61 per 1000) and 1.87 (95% CI, 1.36-2.57 per 1000), respectively. A total of 33 newborns had cyanotic CHD (prevalence 1.63; 95% CI, 1.12-2.28 per 1000). The number and frequency of all significant CHDs is shown in Table 1.

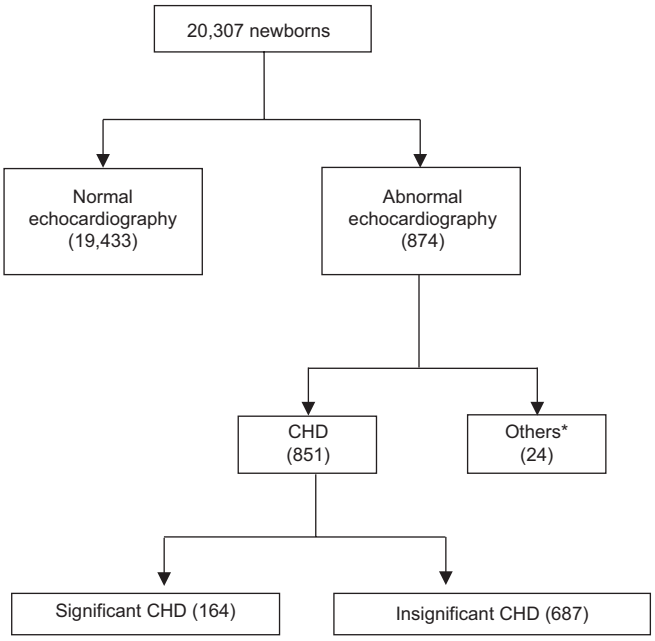

Figure 1: Echocardiographic findings in the total cohort (*n* = 20,307). CHD: Congenital heart disease, Others: Other cardiac findings. *One newborn had congenital heart block (other cardiac finding) along with Tetralogy of Fallot

Twenty-four newborns had abnormal echocardiograms that were not because of CHD. They have been grouped together as other cardiac findings. These are listed along with their birth prevalence in Table 2.

Role of initial evaluation in detection of significant CHDs

Clinical examination results were available for 19,433 newborns (95.7% of the total cohort), out of which 163 had significant CHDs. An abnormal clinical examination was recorded in 552 newborns and 40 of these were true positives. Thus, the sensitivity and specificity of clinical examination were 24.54% (95% CI, 18.15-31.88) and 97.34% (95% CI, 97.11-97.57), respectively.

Pulse oximetry results were available for 19,009 newborns (93.6% of the total cohort). Oxygen saturation was persistently below 95% in 6048 babies; 159 of 19,009 newborns had significant CHDs and oximetry

Table 1: Birth prevalence of significant congenital heart diseases

Congenital heart disease	Number	Significant CHDs (%)
Minor CHD	93	56.7
Restrictive ventricular septal defect	72	43.9
Atrial septal defect	12	7.3
Patent ductus arteriosus	7	4.3
Pulmonary stenosis	2	1.2
Major acyanotic CHD	38	23.2
Nonrestrictive ventricular septal defect	34	20.7
Coarctation of aorta (nonduct dependent)	2	1.2
Aortic stenosis	2	1.2
Major cyanotic CHD	33	20.1
Transposition of great arteries	7	4.3
Hypoplastic left heart syndrome	5	3
Atriovetricular septal defect, pulmonary atresia/stenosis	3	1.8
Tetralogy of Fallot	3	1.8
Double outlet right ventricle	2	1.2
Ebstein's anomaly	2	1.2
Hypoplastic right ventricle	2	1.2
Persistent truncus arteriosus	2	1.2
Single ventricle	2	1.2
Total anomalous pulmonary venous connection	2	1.2
Interrupted aortic arch	1	0.6
Pulmonary atresia, ventricular septal defect	1	0.6
Corrected transposition of great arteries, ventricular septal defect, pulmonary stenosis	1	0.6

Table 2: Prevalence of other cardiac anomalies

Diagnosis	Number
Persistent pulmonary hypertension of newborn	17
Complete heart block	3
Situs inversus dextrocardia with no structural heart disease	2
Supraventricular tachycardia	1
Cardiac tumor	1

detected a saturation <95% in 75 of these. The sensitivity and specificity of pulse oximetry were 47.17% (95% CI, 39.21-55.23) and 68.31% (95% CI, 67.64-68.98), respectively.

DISCUSSION

Previous studies from India have reported a variable prevalence of CHD. The prevalence rate varies from as low as 1.3 per 1000 to as high as 13.28 per 1000 children.[7,13-18] Many of these studies have used the prevalence of CHDs in school children and these are mainly offshoots of prevalence studies for rheumatic fever and rheumatic heart disease. As a large number of congenital heart defects are critical and lead to death in early life itself, these studies on school children have limited value and underestimate the true burden of CHD in India.

The birth prevalence of significant CHDs in our study was 8.07 per 1000 live births. This is almost the same as the worldwide birth prevalence rate of CHDs.[4] Minor CHDs consisted of 56.7% of the significant CHDs and the birth prevalence rate was 4.58 per 1000 live births. Major CHDs consisted of the remaining 43.3% with a birth prevalence rate of 3.49 per 1000 live births. These were further divided into major acyanotic (prevalence 1.87/1000) and major cyanotic CHDs (prevalence 1.63/1000).

VSDs (restrictive and nonrestrictive) were the most common significant CHDs (43.9% and 20.7%, respectively) with a prevalence rate of 5.22 per 1000 live births. ASDs and PDAs consisted of 7.3% and 4.3% of the significant CHDs, with a prevalence rate of 0.59/1000 and 0.34/1000, respectively. This number is lower as compared with the other studies because most of the ASDs and PDAs detected in our study were not large enough to be classified as significant CHDs.[7,13-18] There were two cases each of pulmonary stenosis, coarctation of aorta, and aortic stenosis.

Among the major cyanotic CHDs, transposition of great arteries and hypoplastic left heart syndrome were the most common (seven and five cases; prevalence 0.34/1000 and 0.25/1000, respectively). There were three cases each of atrioventricular septal defects with pulmonary atresia and Tetralogy of Fallot (0.15/1000). The other cyanotic CHDs are listed in Table 1.

In addition, echocardiography in newborns helped identify 17 cases of persistent pulmonary hypertension of newborns, four cases of arrhythmias, and two with situs inversus along with dextrocardia. One case had a cardiac tumor (likely rhabdomyoma).

Initial evaluation of newborns using clinical examination and pulse oximetry was helpful in detecting a few cases of significant CHDs before being diagnosed with echocardiography. Clinical examination detected only 40 of 163 significant CHDs (sensitivity 24.54%) and a persistent saturation of less than 95% on pulse oximetry was seen in 75 of 159 significant CHDs (sensitivity 47.17%). This is less as compared with similar recent studies from other countries.[19-22]

Limitations

Although echocardiography with Doppler is the gold standard for diagnosing CHDs, it is an operator-dependent procedure and hence expertise in performing and interpreting echocardiographic images is very important. Also echocardiography has a 5.4% rate of false positives.[23] Many newborns with ASDs and PDAs at birth were classified as having normal echocardiographic findings because of the small lesion size; a follow-up echocardiogram of such newborns is necessary to ascertain the resolution of these lesions.

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Conflicts of interest

AS: Guarantor, conceptualized and designed the study, obtained funding for the project, performed echocardiograms of newborns, reviewed and critically revised the manuscript, and approved the final manuscript as submitted. AM: Data analysis and interpretation, drafted the manuscript, and approved the final manuscript as submitted. MS, SS: Contributed to design of the study, supervised the study at primary hospital, and approved the final manuscript as submitted. MK: Contributed to design of the study, data analysis, and approved the final manuscript as submitted. RJ, SR: Contributed to data analysis, and approved the final manuscript as submitted. None of the authors have any competing interests.

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