Original Research Article

Screening for congenital heart disease in newborns at urban community health centre

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ABSTRACT

Background: Congenital heart disease (CHD) is a defect in the cardiovascular structure and function and represents a heterogeneous group of defects with little known cause. Most of them are diagnosed in newborn period, yet some may be missed only to be diagnosed later. CHDs present with little or no symptoms and hence are under diagnosed in centres with inadequate facilities. So, this study was conducted to find the prevalence of CHDs in neonates delivered in our centre.

Methods: The study was conducted in an urban community centre from May 2018 to January 2019. Babies delivered here were subjected to clinical examination, pulse oximetry at our centre and detailed ECHO examination was done at a nearby PHC where a trained cardiologist was available.

Results: There were 783 babies delivered during the study period, of which 436 babies underwent echo and formed the study group. Totally 35 (8%) babies had some abnormality on echo. Clinically murmurs were present in 10 newborns. Critical congenital heart disease was seen in 3(0.6%) babies. These babies were diagnosed with severe PS, Bicuspid aortic valve with severe AS and pulmonary atresia. VSD was the commonest malformation followed by ASD and PDA.

Conclusions: Screening of CHD helps to detect defects which otherwise would have been missed. Critical CHD could be diagnosed which enabled us to refer the babies for early surgery.

Keywords: Congenital heart disease, Echocardiography, Newborns

INTRODUCTION

Heart defects are the most common congenital malformations, with an incidence of approximately eight per 1000 live births, while the incidence of critical congenital heart disease (CCHD) is one or two per 1000 newborn babies. In general, CCHD is defined as congenital heart disease (CHD) that leads to death or requires surgery or catheter intervention within 28 days of life. Delayed detection of CCHD increases the risk of acute cardiovascular collapse and death, and is associated with worse outcomes of interventions. As the preference for early discharge after delivery is becoming more prevalent, newborns with CCHD are more likely to develop symptoms at home rather than during their stay in the newborn nursery. Screening for CCHD has previously relied on prenatal ultrasound and postnatal clinical examination, but both such approaches are known to have a relatively low detection rate. It is estimated that up to a third of newborns may be discharged with undiagnosed critical
defects. For more than a decade, many studies have assessed the usefulness of pulse oximetry for improving the detection of CCHD in newborns before hospital discharge (typically, in the newborn nursery); evidence from such studies indicates that pulse oximetry screening is highly effective in detecting CCHD in newborns with hypoxemia but not in those without hypoxemia.\textsuperscript{3,6-8} Echocardiography, especially when performed by pediatric cardiologists, is commonly used for diagnosing CHD, and thus may be helpful in detecting CCHD in newborns earlier and with higher sensitivity than when using other screening methods.

The congenital heart diseases are not fixed anatomic defects that appear at birth but are instead a dynamic group of anomalies that originates in fetal life and the diagnosis is established by 1 week of age in 40-50\% of patients. Most congenital defects are well tolerated in the fetus because of the parallel nature of fetal circulation. It is only after when the fetal pathways are closed the full hemodynamic impact of the abnormality becomes apparent.\textsuperscript{9}

Depending upon the severity, CHD (Congenital Heart Disease) presenting at birth can be categorized into 3 groups - mild, moderate and severe.

Severe CHD includes all cyanotic lesions as well as acyanotic lesions like Large VSD (Ventricular Septal Defect), Large PDA (Patent Ductus Arteriosus), Critical AS (Aortic Stenosis), Critical PS (Pulmonary Stenosis), Critical Coarctation and AVSD (Atrio Ventricular Septal Defect) which require intervention early in life.

Moderate CHD include mild-moderate AS or PS, non-critical coarctation, Large ASD (Atrial Septal Defect) and these are ones that require expert care, but less intensive compared to severe CHD.

Mild CHD (small VSD, PDA, ASD, mild AS or PS) are asymptomatic and often undergo spontaneous resolution.\textsuperscript{10}

Signs and symptoms of severe heart disease in the newborn period include cyanosis, discrepant pulses and blood pressures, congestive heart failure, and cardiogenic shock. Early recognition of congenital heart disease during neonatal life is important as its appropriate and timely management can result in good outcome.

Objectives of the present study were to screen newborns delivered in urban community health centre for congenital heart diseases using pulse oximetry, clinical examination and echocardiogram.

**METHODS**

The study was done prospectively in an urban community health centre at Chennai. Study population were all newborns who underwent echocardiography. The study period was May 2018 to January 2019. The study design was prospective descriptive study.

**Inclusion criteria**

All newborns delivered in community health centre.

**Exclusion criteria**

Outborn deliveries and the babies delivered at urban community health centre at Chennai, but not subjected to ECHO or died before ECHO evaluation.

Permission from institutional ethics committee and well-informed parental consent was taken.

Data was recorded with respect to sex, gestational age, birth weight, maternal risk factors and presenting features. All babies delivered in the centre were subjected to pulse oximetry examination at birth, 24 hrs, 48 hrs and before discharge.

After discharge, the parents were advised to take the child to another primary health centre within 28 days of birth to undergo detailed echocardiography examination by a trained cardiologist.

**Screening protocol**

Authors used the Radical-7 pulse oximeter with the reusable probe (Masimo, Irvine, CA, USA). These devices have low intraobserver and interobserver variability and produce accurate saturations that are stable in active individuals and in low perfusion states, making them suitable for use in the first few hours of a newborn baby’s life.

Authors measured functional saturations in the right hand and either foot in a non-specified order while the baby was still in hospital. The sensor was secured around the palm of the baby’s hand and sole of the foot with a bespoke disposable wrap. Staff nurses well trained in newborn care undertook the tests. Masking of antenatal findings (e.g., suspected abnormalities of cardiac anatomy) was not undertaken because pulse oximetry is an objective test and the reading is unlikely to be affected by knowledge of such findings.

A saturation of less than 95\% in either limb or a difference of more than 2\% between the limb saturation readings (if both were ≥95\%) was judged to be abnormal. These threshold values were chosen to try to increase the sensitivity for detection of left heart obstructive lesions, treatable disorders that were missed most often in studies with higher thresholds. Clinical examination was expedited if an abnormal test result was obtained. If this examination was unremarkable, pulse oximetry was repeated 1-2 hr later. If abnormalities of the cardiovascular system were detected with expedited examination, or saturations remained abnormal during a
second test, the newborn babies were classified as test positive.

**Echocardiography**

The newborns underwent echocardiographic screening within 28 days of life at a nearby urban primary health centre after discharge from our centre. The echocardiographic examination was performed by well experienced pediatric cardiologists, using the same echocardiography machine (Vivid GE Healthcare, Tokyo, Japan) with a 12-MHz transducer.

The examination protocol included two-dimensional and color Doppler imaging in the parasternal, suprasternal, subxiphoid, apical, and, when necessary, modified views. All echocardiographic examinations were recorded and reviewed by another pediatric cardiologist, who served as a second observer blinded to the patient identity. Sedation was not required in any newborn because echocardiographic screening was performed during natural sleep.

Atrial septal defect was defined as the presence of an intra-atrial communication ≥5 mm in diameter, with an enlarged right atrium and right ventricle. An intra-atrial defect ≤4 mm in size at the fossa ovalis was considered to represent patent foramen ovale.

**RESULTS**

The study was done at urban community health centre at Kodambakkam and echocardiography of recruited cases were done at urban primary health centre at Saidapet, Chennai.

A total of 783 newborns were delivered at our centre during study period. All the babies were subjected to pulse oximetry examination at birth and 1 to 2 hours after birth, at 24 hours and before discharge. Of 783 babies, except for 2 babies, rest passed pulse oximetry test. Of the 2 babies which failed pulse oximetry, one was referred to tertiary care hospital where it died before echocardiographic evaluation and was excluded from the study group.

Detailed physical examination and cardiovascular examination was done by a paediatrician at the centre. Babies were assessed for central cyanosis, abnormal precordial pulsations, evidences of shock, congestive cardiac failure and presence of murmurs. Of 783 babies central cyanosis was present in 2 babies, shock in 1 and murmurs were detected in 10 babies.

All the parents who delivered here were advised to take their babies to nearby urban primary health centre at Saidapet for detailed echocardiographic examination by a cardiologist. Of 783 babies delivered during the study period 436 (55.6%) babies underwent echocardiography and they formed the study population.

**Characteristics of study population**

With respect to gender males and females were equally divided. Total 433 (99.3%) babies were term babies and only 3 babies were preterm.

With respect to birth weight most of them were appropriate for gestational age 400 (91.7%), while 31 (7.1%) were small for gestational age, only 5 (1.2%) were large for gestational age (Table 1).

**Table 1: Age, gestational age, weight distribution of study population.**

| Characteristics       | No. | Percentage |
|-----------------------|-----|------------|
| Gender                |     |            |
| Male                  | 221 | 50.6       |
| Female                | 215 | 49.4       |
| Gestational age       |     |            |
| Term                  | 433 | 99.3       |
| Pre-term              | 3   | 0.7        |
| Post-term             | -   | -          |
| Birth weight          |     |            |
| AGA                   | 400 | 91.7       |
| SGA                   | 31  | 7.1        |
| LGA                   | 5   | 1.2        |

**Characteristics of babies with abnormal echo**

Total 35 (8%) babies from our study population had abnormal echo findings with a female preponderance. 26 (74.3%) babies were females while 9 (25.7%) only were males. Most of the babies were term babies. Only one preterm had abnormality.

Although majority were with normal weight (80%) still a significant number of babies with abnormal findings belonged to small for gestational age (20%). None belonged to LGA (Table 2).

**Table 2: Characteristics of babies with abnormal echo.**

| Characteristics       | No. | Percentage |
|-----------------------|-----|------------|
| Gender                |     |            |
| Male                  | 9   | 25.7       |
| Female                | 26  | 74.3       |
| Gestational age       |     |            |
| Term                  | 34  | 97.1       |
| Pre-term              | 1   | 2.9        |
| Post-term             | -   | -          |
| Birth weight          |     |            |
| AGA                   | 28  | 80         |
| SGA                   | 7   | 20         |
| LGA                   | 0   | 0          |

**Types of malformations**

**Cyanotic heart disease**

Only one among 35 had a cyanotic heart disease. Child had multiple malformations including PA (Pulmonary atresia). Large PDA Large conotruncal VSD, small PFO.
A cyanotic heart disease

VSD was the commonest malformation seen in 11 (31.4%) and almost equally divided between males and females followed by ASD which was seen in 6 (17.2%) babies. Atrial septal defect (ASD) was predominantly seen in females (5/6). PFO a normal variant was seen in 6 (17.2%) babies.

Patent ductus arteriosus (PDA) was seen in 3 babies. Multiple lesions were seen in 2 babies. One male child had VSD+PFO and a female child had ASD+ small main pulmonary artery. LSVC which is a functionless abnormality and which becomes more important during surgeries were seen in 2 babies.

Valvular stenotic lesions

One child had valvular pulmonary stenosis and underwent valvuloplasty in newborn period itself and another child had bicuspid aortic valve with severe aortic stenosis (Table 3).

Table 3: Types of malformations on echo.

| Malformations | Male | Female | Total |
|---------------|------|--------|-------|
| VSD           | 5 (14.3%) | 6 (17.1%) | 11 (31.4%) |
| PFO           | 3 (8.6%) | 3 (8.6%) | 6 (17.2%) |
| ASD           | 1 (2.8%) | 5 (14.4%) | 6 (17.2%) |
| Aortic stenosis | - | 1 (2.8%) | 1 (2.8%) |
| Pulmonary stenosis | - | 1 (2.8%) | 1 (2.8%) |
| PDA           | 1 (2.8%) | 4 (11.8%) | 5 (13.6%) |
| LSVC          | 2 (5.6%) | 2 (5.6%) | |
| Multiple malformations | | | |
| VSD + PFO     | 1 (2.8%) | - | 1 (2.8%) |
| ASD + small main pulmonary artery | - | 1 (2.8%) | 1 (2.8%) |
| Pulmonary atresia large Conotruncal VSD /MAPCAS/ large PDA/ small PFO | | 1 (2.8%) | 1 (2.8%) |

Murmurs and CHD

Clinically murmurs were detected in 10 (28.6%) newborns. Three babies with PFO had systolic muemurs and 2 babies with PDA had murmurs.

Each baby with PS, AS and pulmonary atresia also had murmurs on auscultation. None of the babies with ASD, VSD had murmurs. 2 babies who had normal echo also had systolic murmurs (Table 4).

Classification of CHD

CHDs were classified into various categories as severe, moderate and mild. One baby with severe pulmonary stenosis, one with bicuspid aortic valve with severe aortic stenosis and one baby with pulmonary atresia/conotruncal VSD/MAPCAS were classified under severe CHDs.

ASD lesions with a defect in atrial septum with a diameter of 5 mm or more were classified under moderate CHDs. As such 7 (20%) babies come under this category. Babies with tiny VSDs and tiny PDAs were classified under mild category, 17 (48.8%) babies came under mild CHDs. Normal variants were those with PFO 6(17.1%) and LSVC 2 (5.6%). They together formed 22.7% of babies (Table 5).

Table 4: Murmurs and CHD.

| Echo finding | Present | Absent | Percentage |
|--------------|---------|--------|------------|
| PFO          | 3       | 3      | 50         |
| PDA          | 2       | 3      | 40         |
| PS           | 1       | 0      | 100        |
| AS           | 1       | 0      | 100        |
| PA/VSD/MAPCAS| 1      | 0      | 100        |
| VSD          | 6       | 0      |            |
| ASD          | 6       | 0      |            |
| LSVC         | 2       | 0      |            |
| NORMAL       | 2       |        |            |

Table 5: Severity of CHD.

| Severe/ critical | Number | Percentage |
|------------------|--------|------------|
| PS               | 1      | 2.8        |
| AS               | 1      | 2.8        |
| Pulmonary atresia/ VSD | 1 | 2.8 |

Moderate

| Severity | Number | Percentage |
|----------|--------|------------|
| ASD      | 6+1=7  | 20         |
| VSD      | 11+1=12| 34.3       |
| PDA      | 5      | 14.3       |

Normal variants

| Number | Percentage |
|--------|------------|
| PFO    | 6          | 17.1       |
| LSVC   | 2          | 5.6        |

Outcome

As on date one child with severe pulmonary stenosis underwent valvuloplasty. One baby with congenital bicuspid valve with severe aortic stenosis and one with pulmonary atresia have been wait listed for surgery. Rest of the babies in moderate and mild CHDs categories are alive and thriving well.

DISCUSSION

Congenital heart disease (CHD) is the most common congenital abnormality found in paediatrics comprising...
approximately 25% of all the congenital anomalies. The objective of present study is to screen all the newborns delivered in our centres for CHD by clinical examination, pulse oximetry and ECHO evaluation. 783 neonates were delivered in our centre. ECHO was done in 436 (55.5%) babies.

Incidence of CHD in our studies was 8% which was comparable to the study done by Vaidyanathan et al. (7.3%).11 The overall prevalence of CHD reported in this study is higher than that reported previously from various population based studies. This is accountable by the detection of minor, self-resolving lesions due to use of echocardiography for screening all babies. Though murmur, cyanosis and abnormal pulse oximetry were identified as predictors of CHD, the sensitivity of these signs to detect CHD were very low (combined clinical evaluation and pulse oximetry were positive in 22.8% of babies only).

There was a female preponderance in present study. 72.7% were females. But a study done by Rashid et al, had reported a slight male preponderance.12 VSD was the commonest malformation seen in 34.3% of cases, which was comparable to study done by Rashid et al.12 This was followed by ASD which was seen in about 20% of cases, this was also comparable to the study done by Rashid et al. (14%).12 Next common malformation was PDA seen in 14.3% as against 18% in the study done by Rashid et al.12 Cystotic heart disease was seen in one patient (2.8%) whereas Rashid et al, reported (29%).12 There was 1 case of pulmonary atresia whereas Rashid et al, had reported TOF followed by TGA.12

Critical CHD - valve stenosis is the fourth most common acyanotic congenital heart lesion (5.6%). Severe Pulmonary stenosis was seen in 1 (2.8%) in this study as against 0.66% reported by Rashid et al.12

Bicuspid aortic valve with stenosis was also seen in 2.8%. Abnormal clinical finding especially murmurs are seen in 22.8% of the babies which is comparable to study done by Vaidyanathan et al, (20%).11

In present study severe CHD was seen in 8.4%, moderate CHD in 20% and mild in 48.6% which is comparable to study done by Pillai et al, (severe CHD - 13.9%, moderate CHD - 20.8%, mild CHD - 40.3%).13 Normal variants were seen in 25.7% in current study and are comparable to 25% seen in study done by Pillai et al.13

CONCLUSION

Echocardiographic screening is useful in detecting CHDs in neonates. Incidence of CHDs is higher because of detection of normal variants and tiny defects which usually close within a year. Combining pulse oximetry and clinical evaluation is useful in detecting critical congenital heart defects and they are of less importance in detecting moderate and mild CHDs. For the diagnosis or exclusion of CHDs, ECHO evaluation is essential. But since the procedure is cumbersome and expensive routine screening cannot be advocated in higher centres with a higher case load.

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