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ORIGINAL ARTICLE

Foetal Cardiac Anomalies: Experience in a Primary Referral Centre

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Abstract Congenital heart disease (CHD) is one of the most common congenital anomalies reported. Incidence of CHD is 8–9 per 1000 live births in data published from the west. There is very little published data on the incidence of foetal cardiac anomalies in India. We tried to find out incidence and spectrum of foetal cardiac anomalies in second and third trimester during routine ultrasound examination in a primary referral centre. Of 11,760 fetuses in the 2nd and 3rd trimester, 104 were found to have cardiac anomalies with incidence of 8.8 per 1000. Ventricular septal defect was the most common lesion, followed by chamber asymmetries and outlet abnormalities.

Keywords Ultrasound (US) examination · Congenital heart disease (CHD) · Incidence and spectrum · Prenatal · India

Introduction

Congenital heart diseases (CHD) are the most common congenital abnormalities [1]. An incidence of 8–9 per 1000 live births has been reported on large population studies worldwide [1, 2]. The overall incidence of CHDs may be in order of 50 per 1000 live births if all subtle cardiac anomalies are counted, including bicuspid aortic valve, aneurysm of atrial septum and persistent left superior vena cava (PLSVC) [2, 3]. In Asia, estimated prevalence is 9.3 per 1000 live births [1, 2]. There is very little published data available about CHDs in India [2, 3]. The available data shows the prevalence of CHDs in India to vary from 2.25 to 5.2 per 1000 live births [2–5]. No data is available on prenatal incidence of foetal cardiac anomalies in India. Prenatal diagnosis of cardiac anomalies plays crucial role in parental counselling and perinatal management. In the present retrospective study, we tried to find out incidence and spectrum of foetal cardiac anomalies in second and third trimester during routine ultrasound (US) examination in a primary referral centre in India.

Materials and Methods

We retrospectively analyzed the data of 11,760 US examinations of pregnant patients in second and third trimester from 2014 through 2016 in a primary referral centre. All major and minor cardiac anomalies in patients were recorded to find out the incidence and spectrum of the same. Cases with associated extra cardiac anomalies were also included.

Cardiac anomalies were divided into three main groups: (1) four chamber (4CH) abnormalities (2) outlet abnormalities (3) venous and other miscellaneous abnormalities.

Gestational age wise distribution of anomalies was also calculated in following group:- 12–14 weeks, 15–20 weeks, 21–28 weeks and > 28 weeks.

Foetal cardiac evaluation was carried out as per the ISUOG, AIUM, American Society of echocardiography screening guidelines [6–8].

Systematic cardiac examination was done in all second and third trimester cases in cardiac set up using gray scale, M-mode and colour imaging using convex probe (C1-4 MHz). The endocavitatory (TV 4–9 MHz) and linear (L

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5–12) probes were also used if needed. Foetal position preferred was cardiac apex pointing anteriorly and laterally in most of the cases.

Quick cardiac screening was done by taking caudo-cranial sweep from foetal abdomen to the base of neck to look for situs, position and axis of the heart and for any major deviation from normal.

Systematic examination was done by section wise analysis with adequate zooming as per ISUOG, AIUM, American Society of echocardiography screening guidelines [6–8].

Sections documented and studied included 4 CH, foetal upper abdomen, left ventricular outflow tract (LVOT), right ventricular outflow tract (RVOT), three vessel trachea (3VT) and confluence of arches. Other optional sections such as short axis, aortic and pulmonary arches view, longitudinal view and caval view were used whenever necessary. Two dimensional and colour mode examination was done in all sections. Ductus venosus and cord insertion was also documented. Rate and rhythm was evaluated using 'M' mode. Pulse wave evaluation was kept optional for suspected abnormal cases.

Results

We retrospectively analyzed the data of 11,760 US examinations of pregnant patients. Cardiac abnormalities were diagnosed in 104 cases. This gives incidence of 8.8 per 1000 (second and third trimester) cases. Isolated cardiac anomalies were seen in 88 cases (84.6%) and associated extra cardiac abnormalities were seen in 16 cases (15.4%). Majority of the cardiac anomalies were primarily diagnosed on 4CH view (42.3%) and outflow tract view (20.2%), amongst which isolated ventricular septal defect (VSD) (26.9%) is the most common, followed by hypoplastic left heart syndrome (HLHS) (9.6%), arch anomalies (10.5%), tetralogy of Fallots (TOF) (5.8%), transposition of greater arteries (TGA) (3.8%), atrio-ventricular septal defect (AVSD) (1.9%) and tricuspid atresia with hypoplastic right heart syndrome (HRHS) (1.9%) respectively. Other abnormalities (37.5%) included venous, situs, functional and complex anomalies (Tables 1, 2, 3). Table 1 shows the classification of abnormalities.

Table 1 General classification of abnormalities

Cardiac view	No. of cases	Percentage	Per 1000 cases
Abnormal 4 CH view	44	42.3	3.7
Abnormal outlet and arches	21	20.2	1.7
Other abnormalities (miscellaneous, venous and situs)	39	37.5	3.3

Table 2 Gestational week wise distribution of cardiac defects

Gestational weeks	No. of cases	Percentage	Per 1000 cases
12–14	10	9.7	0.8
15–20	37	35.6	3.2
21–28	29	27.8	2.4
> 28	28	26.9	2.3

Table 3 Spectrum of specific cardiac anomalies

Abnormality	No. of cases	Percentage	Per 1000 cases
Isolated VSD	28	26.9	2.4
ASD	2	1.9	0.15
AVSD	2	1.9	0.15
HLHS	10	9.6	0.8
Tricuspid atresia (HRHS)	2	1.9	0.2
TGA	4	3.8	0.3
TOF	6	5.8	0.5
Arch anomalies	11	10.5	0.9
Venous	8	7.7	0.7
Situs abnormalities	10	9.7	0.8
Other (minor, tumors and functional)	21	20.3	1.8

VSD ventricular septal defect, *ASD* atrial septal defect, *AVSD* atrioventricular septal defect, *HRHS* hypoplastic right heart syndrome, *TGA* transposition of greater arteries, *TOF* tetralogy of Fallot

Table 2 lists the cardiac anomalies according to the gestation in which they were detected.

Discussion

There is very little published data available till date on incidence of CHD specific to Indian population [2, 3]. There is no reported data available on prenatal incidence of cardiac anomalies in India. This study was done to assess the prenatal incidence and spectrum of cardiac anomalies in Indian population referred for routine US examination in a primary referral centre. From our analysis, the incidence of CHD is 8.8 per 1000 in second and third trimester prenatal US examination. If a systematic basic protocol of cardiac screening is followed, almost all significant anomalies can be detected in prenatal examination, except for minor atrial septal defect (ASD) and patent ductus arteriosus (PDA).

We found majority of the cardiac anomalies were primarily diagnosed on 4CH view (42.3%), amongst which isolated VSD (26.9%) is most common followed by HLHS (9.6%), AVSD (1.9%) and HRHS (1.9%) respectively. Most of the VSDs were minor muscular and were picked up easily on colour flow examination, especially in late

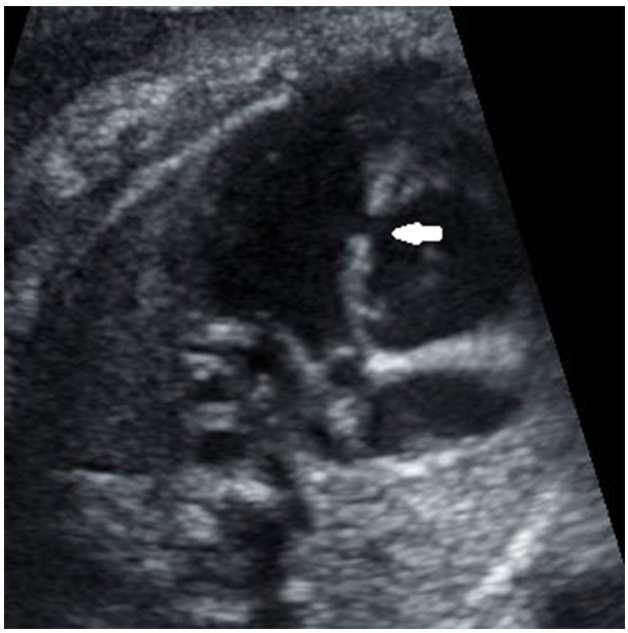

Fig. 1 VSD, ventricular septal defect, a small opening in muscular portion of inter ventricular septum (arrow)

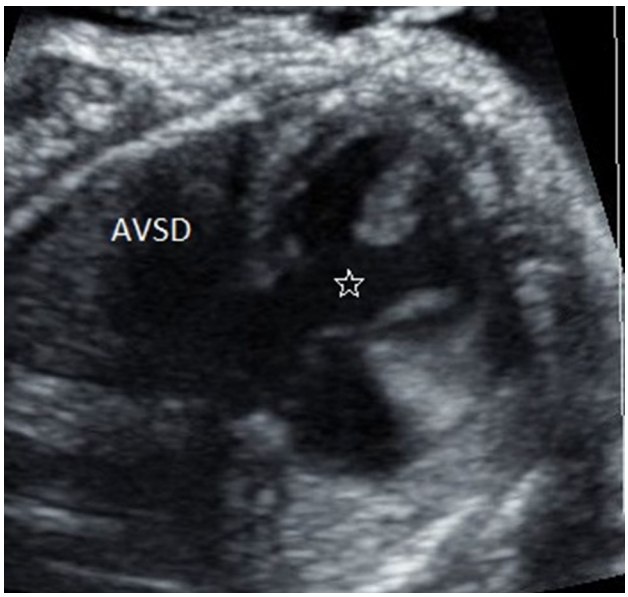

Fig. 2 4CH view-AVSD, atrioventricular septal defect, absent crux with single atrioventricular valve (star)

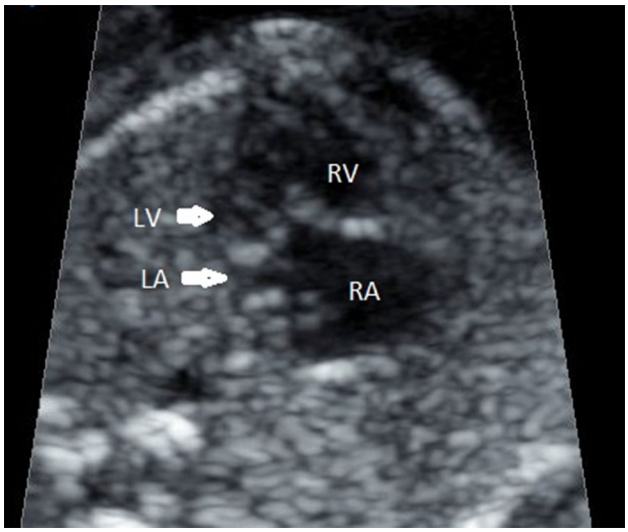

Fig. 3 4CH view-HLHS, hypoplastic left heart syndrome, significantly small left ventricle and atrium (arrow)

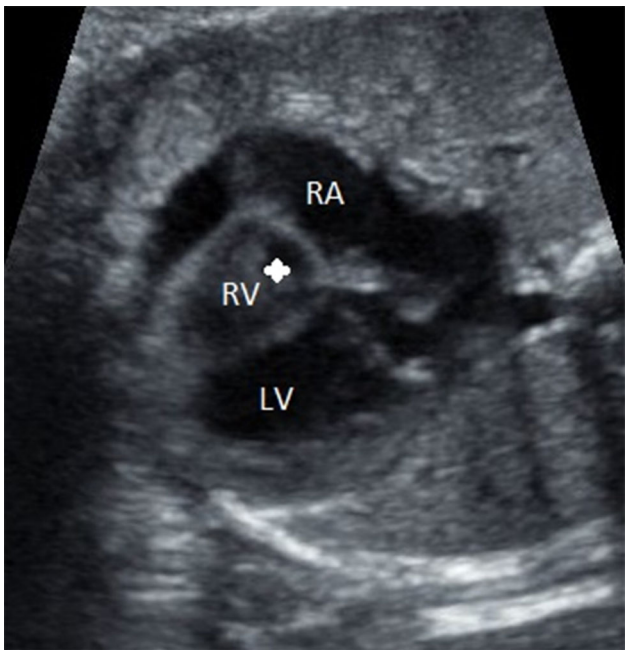

Fig. 4 4CH view-HRHS, hypoplastic right heart syndrome, small right ventricle with thick tricuspid valve (star)

second and third trimester (Fig. 1). Other VSDs were larger muscular and membranous types which were associated with other cardiac anomalies (Fig. 2). Most of HLHS were associated with mitral and aortic valve atresia and HRHS cases were associated with tricuspid atresia (Figs. 3, 4). One case of Ebstein's anomaly was diagnosed at around 17 weeks.

Cases related to abnormal origin, size and misalignment of outlets were (20.2%), which comprised TOF, TGA and double outlet right ventricle (DORV) and arch anomalies such as suspected coarct, right aortic arch etc (Figs. 5, 6).

Other anomalies (37.5%) included venous anomalies like PLSVC, total anomalous pulmonary venous drainage (TAPVC), absent ductus venosus (DV), interrupted inferior vena cava, situs abnormalities, functional abnormities and cardiac tumor, etc. There were few complex cases which had more than one cardiac abnormality.

Cases associated with extra cardiac anomalies were (15.4%), such as central nervous system, spinal, limb

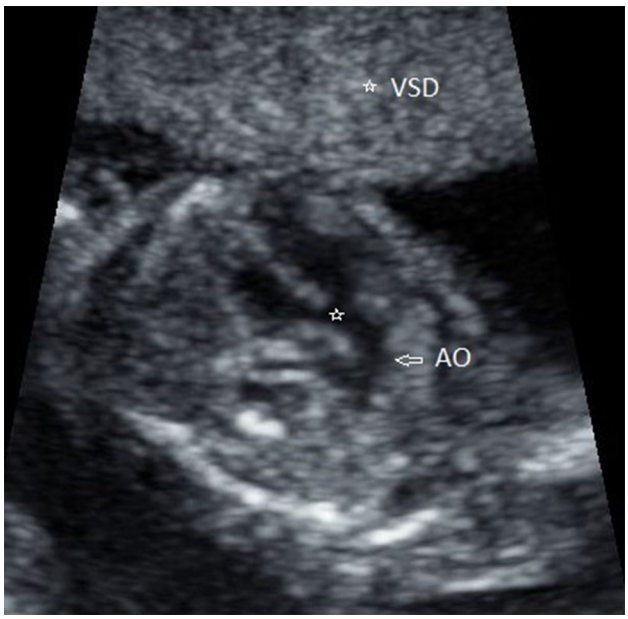

Fig. 5 Left ventricular outflow tract view-TOF, tetralogy of Fallot, membranous VSD (star) with overriding of aorta (arrow)

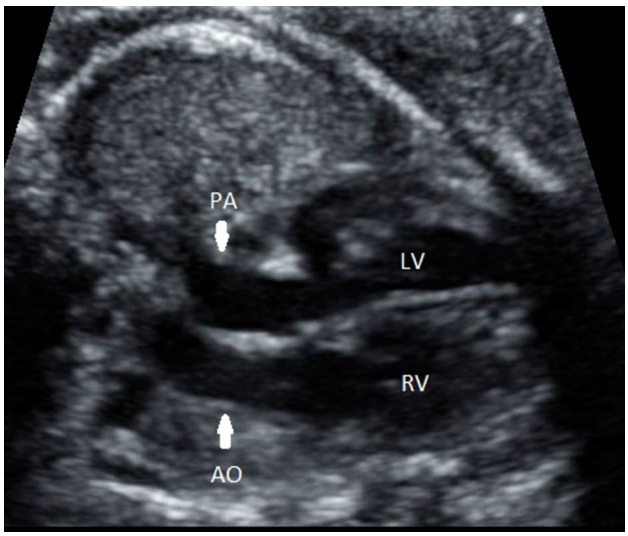

Fig. 6 Outlet view-TGA, transposition of greater arteries, parallel aorta and pulmonary artery (arrows)

anomalies, two vessel cord and abnormal nuchal translucency (NT) etc.

We found that anomaly detection rate is equal in early second, late second and third trimester, hence systemic cardiac evaluation is crucial in every scan irrespective of the stage of pregnancy.

In the present study, we could diagnose most of the spectrum of cardiac anomalies in prenatal examination. The findings were in concordance with most of the international published data on CHD.

Though CHD is the commonest prenatal anomaly [1–4], it is largely unrecognized and underestimated in India [3]. This is mainly because of lack of awareness and unavailability of uniform prenatal and postnatal diagnostic facilities. Knowing prenatal incidence and exact spectrum of cardiac anomalies is very crucial in parental counselling, obstetric management and multidisciplinary care. Therapeutic terminations can be advised before 20 weeks if the anomalies are complex. If CHD is associated with other abnormalities then chromosomal abnormalities can be suspected. If the repairable abnormalities go undiagnosed, they have great impact on infant mortality and morbidity. Our study has shown that incidence in Indian population is comparable with incidence reported from rest of the world. The study also shows importance of systematic approach to foetal cardiac evaluation. Hence, our study will definitely be helpful to imaging specialists and clinicians in India to improve the prenatal diagnosis of CHDs.

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