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Original Research Paper

Pathology

Congenital Malformations Of Heart In Perinatal Autopsies.

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ABSTRACT

BACKGROUND Congenital malformations remain a common cause of perinatal deaths and even though ultrasonogram can give fairly accurate diagnosis, perinatal autopsy is essential to confirm the diagnosis and look for associated malformations. **OBJECTIVES** To emphasize the importance of perinatal autopsy in diagnosing congenital malformations of heart and to study the distribution of congenital malformations of heart in perinatal deaths. **MATERIALS AND METHODS** The present study comprised 100 hearts from consecutive perinatal autopsies conducted in Department of Pathology, M.R Medical College, Gulbarga, over a period of 1year. The protocol included the removal of thoracic, cervical, abdominal and pelvic organs en block and subsequently dissected into organ blocks. Histological sections were taken from heart, lung, liver, kidney, thymus, brain, placenta and umbilical cord. **RESULTS** Out of 100 cases, 05 cases (5%) showed congenital malformations of heart with M : F 1.5:1. Majority of cases with heart malformations were IUDs (60%) with maximum 03 cases between 30-34 weeks; mean Gestational age 31weeks and mean gestational weight between 2000grams. Most common congenital malformation was VSD (3 cases) followed by one case each of ASD and TGA. 01 case (ASD) presented as part of Edward's syndrome. **CONCLUSION** This study highlights the importance of perinatal autopsy in confirming the diagnosis of congenital anomalies by prenatal ultrasound findings.

KEYWORDS : Perinatal autopsy, congenital malformations, prenatal ultrasound

INTRODUCTION:

With an incidence of up to 5%, congenital cardiovascular malformations are among the most prevalent birth defects and are the most common type of pediatric heart disease. Because of their poor prognosis they contribute significantly to infant mortality. Approximately 1% of individuals have significant forms of congenital heart disease that are diagnosed in the first year of life[1].

Various epidemiological studies have shown that in liveborn infants the incidence of congenital heart defects is between 04-08 per 1000,in stillborn infants the incidence is 10 times that of live births[2].

The prenatal diagnosis of congenital heart defects is often difficult and can only be carried out competently at specialist centers. The main aim of this study was to emphasize the importance of perinatal autopsy in diagnosing congenital malformations of heart and to study the distribution of congenital malformations of heart in perinatal deaths.

MATERIALS ANDMETHODS:

The present autopsy study was conducted in the department of Pathology in a tertiary care centre, over a period of one year. Consecutive autopsies were carried out during the study period & were included in the study. Detail clinical history regarding age, sex, cause of death was noted. USG findings were recorded and correlated wherever possible.

All the hearts with aorta were fixed in 10% formalin. Thorough morphological examination of heart was carried out. Relation of the great vessels was noted, any variations recorded. The gross photographs of the lesions were taken to demonstrate the defect present. The heart was then cut open along the flow of blood as described by Virchow (Inflow – outflow technique).

Consent was taken from Institutional Ethics Committee prior to commencement of study.

RESULTS:

Total 100 autopsies were performed during study period. Congenital heart diseases were found in 05 cases.

Table 1 : Age and Sex distribution of autopsied cases with congenital malformations of heart

Gestational age	Male	Female	Total	
	No.	No.	No.	%
20-24 wks	0	0	0	0
25-29 wks	1	0	01	20
30-34 wks	2	1	03	60
35-39 wks	0	1	01	20
TOTAL	03	02	05	100

Table 2 : Mode of death in autopsied cases with congenital malformations of heart

Mode of death	No. of cases	Percentage
Abortion (spontaneous)	02	40
Termination (therapeutic abortion)	0	0
IUD	03	60
Total	05	100

Table 3: Showing birth weight distribution in perinatal cases with congenital malformations of heart

Birth weight (grams)	No. of cases	Percentage
400-1000	00	00
1001-2000	03	60
2001-3000	01	20
3001-4000	01	20
TOTAL	05	100

Table 4: Showing congenital anomalies of heart in perinatal autopsies

Congenital heart disease	No. of cases
VSD	03
ASD	01

TGA	01
Total	05

Maximum number of cases noted were of Ventricular septal defect VSD (03 cases), one case of Atrial septal defect (ASD) and one case of Transposition of great arteries (TGA) (Table no. 4). VSD and TGA were noted as isolated defects. One case of ASD, presented as part of Edward's syndrome. Fetus with Edward's syndrome was a male, of the gestational age 28weeks weighing 1500 grams. There was slight male preponderance in the incidence of congenital heart defects in the present study (M: F = 1.5: 1). Most common gestational age with congenital malformations of heart was 30-34 weeks with mean gestational age of 31weeks(Table no.1). Out of 05 cases, 03 cases had gestational weight between 1000-2000 grams with mean gestational weight of 2000 grams(Table no. 3). Of the 05 cases of congenital heart diseases, Intrauterine death (IUD) was the mode of death in 03 cases and 2 were abortion(Table no.2).

DISCUSSION:

The incidence of congenital heart disease in 100 cases presenting for autopsy during the period of this study was 5% (05 cases). The incidence of congenital heart defects in our study correlated well with the study by Kaiser *et al*, i.e., 4.6%[3]. In the study by Khiste *et al*, which included 270 autopsies, 10 cases of congenital malformations were noted with an incidence of 2.7%[4]. Richter *et al* reported the incidence of congenital heart disease to be 26.5% in their autopsy study[5].

On the other hand, Tennstedt *et al* reported congenital heart disease in 16% of autopsies i.e., congenital heart defects were found in 129 cases out of 815 autopsies[2]. In the study by Padma *et al*, 102 autopsies were studied which included congenital anomalies from all the systems and they noted only a single case of heart defect i.e., Dextrocardia[6]. In the study by Prabhala *et al*, 23 autopsies were studied which found one case of congenital heart defect (4.3%)[7].

The incidence of congenital heart disease, according to different studies show wide variation.

In our present study, Ventricular septal defect (VSD) was the most common defect noted (60%) followed by ASD and TGA(single case each) (Fig 1 & Fig.2). Khiste *et al* found VSD as the most common defect (30%) followed by ASD (20%) and pulmonary stenosis (20%)[4]. Tennstedt *et al* also reported VSD(28%) as the most common defect in their study followed by atrioventricularseptal defect (AVSD) (16%) and hypoplastic left heart (HLH) (16%)[2]. Ventricular septal defect has been reported to be the most frequent cardiac defect in various other autopsy studies.

VSD and TGA, in our study occurred as an isolated defects. ASD was seen as part of Edwards syndrome. Other features in the fetus with Edwards syndrome apart from ASD were cleft lip, cleft palate, low set ears(Fig.2).

In our study, one case (20%) was associated with extracardiac malformations (Edwards syndrome). In the study by Tennstedt *et al*, 11 cases showed isolated heart defect (no other cardiovascular or extracardiac malformations present)[2]. In 66% of the cases further cardiovascular anomalies were observed. For example, ventricular septal defect occurred together with double outlet right ventricle, coarctation of the aorta, tetralogy of Fallot. 85 cases (66%) were associated with additional extracardiac malformations.

In contrast, in the Baltimore-Washington Infant Study (BWIS) only 27% of the congenital heart defects were associated with extracardiac anomalies, the most frequent being of the CNS, eyes, Gastrointestinal system, Genitourinary system and abdominal[8]. In BWIS majority of cases of TGA and right and left sided obstructive defects were not associated with extracardiac anomalies[8]. For malformations of the outflow tract, Lurie *et al* gave a ratio of isolated extracardiac anomalies to combined extracardiac anomalies of 1:2.5[9].

The sex distribution amongst the fetuses with cardiac malformations was 1.5:1 in the present study. It was slightly higher in males compared to females, which correlates well with various other autopsy studies[4,10].

In our study,03 cases(60%) of deaths in fetuses with congenital heart defects were Intrauterine death (IUD), 02 cases(40%) were spontaneous abortions, whereas in the study by Khiste *et al*, 50% of deaths occurred within 5 days of birth, 30% of deaths occurred between 4 months -1 year of life[4]. Tennstedt *et al* reported 22% induced abortions (99 cases), 9% spontaneous abortions (20 cases) and 7% stillbirths (10 cases)[2]. Hegerty AS *et al* It is estimated that 1/3rd to 2/3rd of children with congenital heart disease die within 1st year of life[11]. According to a study by Campbell M, 17.7% of cases of ventricular septal defect survived beyond the age of 20 years[12].

In the study by Antia AU, most of the deaths (50%) in cases of congenital heart disease occurred in the perinatal period in which internal malformations were not suspected clinically[10].

Thus autopsy is a valuable tool for detecting visceral malformations adding to the clinical diagnosis and counseling the parents for subsequent pregnancies. Knowledge of demographic variations of congenital heart disease may lead to new etiological insights & may be useful for preventive therapies. Socioeconomic status, education, urbanization, climatalogical factors, ethnicity, lifestyle & health care seeking behavior of the patient play an important role in congenital heart disease incidence & mortality[13].

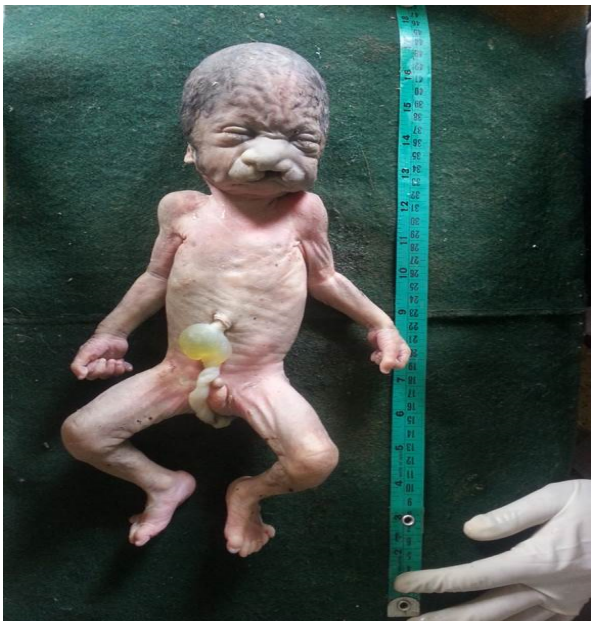
Cardiac development is regulated by complex mechanisms involving interaction between genetic and environmental factors. The etiology of the majority (70-80%) of congenital heart defects is still unexplained. With progress in molecular and developmental biology, our understanding of factors that influence cardiac development is likely to increase. In the last few years it has been shown, for example, that conotruncal heart defects such as TOF, truncus arteriosus communis, double outlet right ventricle, and TGA, together with various types of VSD are associated with a microdeletion on chromosome 22. Raymond *et al* have described five cases with congenital heart defects showing a deletion of 22q11.2 at prenatal diagnosis[14]. The incidence of del(22) is reported to be 1/5000– 10000 births. In 106 fetuses with congenital heart defects and a normal karyotype, Lucy *et al* found two cases with a 22q11.2 deletion[15]. In cases where conventional methods showed a normal karyotype, chromosome abnormalities (7q11.23, 10p13, and 8p deletions) were assumed to be the cause of the cardiac anomalies[15]. In the study by Tennstedt *et al* , one of cases with a ventricular septal defect associated with coarctation of the aorta, it was possible to detect a deletion at chromosome 22 by molecular genetic investigations[2].

Till date aborted fetuses have not been included in investigations of the genetic basis of heart defects.

CONCLUSION:

Congenital malformations are important cause of perinatal death and perinatal autopsies are still the standard criterion for confirming the diagnosis for fetal death. Though prenatal ultrasonogram reasonably predicts the malformations, fetal autopsy is essential to look for additional malformations. This study confirms the utility of fetal autopsy in identifying the cause of fetal loss which will help in genetic counselling of the couple.

Fig. 1 Edwards syndrome
a. Fetus with cleft lip, cleft palate, low set ears.



b. Atrial septal defect (ASD)

Figure 2: Transposition of Great Arteries(TGA)
a- USG- showing main pulmonary artery arising from leftventricle

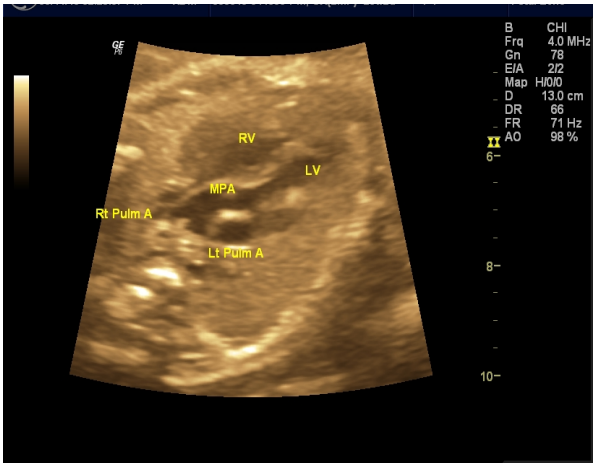
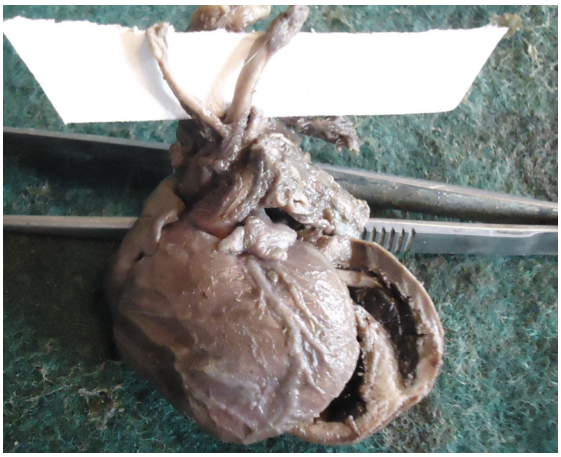


Fig 2b
Gross heart showing anterior aorta and posterior pulmonary artery with division



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