Does fetal echo help the fetus?

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Prenatal diagnosis of fetal abnormalities has come of age. Fetal echocardiography has become a standard prenatal diagnostic modality for the fetus at risk of having cardiac abnormalities. It is expected that early diagnosis of heart lesion in the fetus might help in the subsequent appropriate management of the newborn in the tertiary care center after delivery. Associated non-cardiac abnormalities, anticipated course of the cardiac illness, and parental expectations and attitudes may determine the outcomes of the fetus after the diagnosis of cardiac illness. The value of this investigation, of necessity, will vary with the availability of neonatal care in the society. In India, where less than 10% of neonates with critical cardiac lesion are undergoing operative treatment,[1] the impact of fetal echocardiography may be different than that in the regions with different infrastructure.

The present study by Vaidyanathan et al. in this issue of annals^[2] describes a fetal echocardiography registry and analyzes outcome of the fetuses diagnosed to have congenital heart defects (conotruncal abnormality) in India. The authors need to be congratulated for maintaining such a registry and analyzing the data. Of 1292 fetal echocardiograms performed in 3 years, nearly 25% had a cardiac abnormality; such high yield of fetal echocardiography is because of the referral of fetuses with abnormal scans by the referring physicians. The proportion of conotruncal abnormality (20%) amongst the heart lesions appears higher than that found in the studies in the neonates. This is not surprising since the epidemiology of congenital heart disease (CHD) in the fetus is significantly different from that in the neonates. Much higher incidence of CHD including a higher incidence of complex lesions has been reported in prenatal studies, especially those including abortions and stillborn fetuses. As summarized by Hoffman, much higher occurrence of chromosomal aberrations were also reported in these studies.[3]

Accuracy of fetal cardiac diagnosis was quite high in the present study. The authors have highlighted three aspects of the fetal echocardiographic practice in India: Mainly late referrals, low utilization of tertiary care services despite the diagnosis, and high rates of non-survival in the fetuses. This is important as the study is from Kerala with high human development indices, and the results may be somewhat worse for the fetuses from other parts of the country, although that remains to be studied.

Accurate prenatal cardiac diagnosis should have resulted in increasing utilization of neonatal care services. But it did not happen. In a study from the United Kingdom, the expansion of fetal cardiac services led to increased workload of tertiary pediatric cardiac care units. Some centers report up to 20% of total neonatal admissions due to underlying cardiac defect having had fetal cardiac diagnosis.[4] Instead, in the present study, nearly half of the fetuses with conotruncal defects did not result in fetal survival, possibly because of termination of the pregnancy. Many of the cardiac anomalies seem potentially correctable. It is recognized that the natural history of prenatally diagnosed lesions might differ from that of the same lesion diagnosed postnatally in the neonate. Progression in the anatomic severity of lesion has been well documented in the fetal life, [5] as well as a high incidence of chromosomal defects and associated abnormalities are found in these fetuses with congenital heart defects. [6] Thus, fetal non-survival and termination of pregnancy stem from many causes.

Appropriate counseling was done by the cardiologists, medical social worker, and pediatrician after the fetal echo in the present study. [2] Although a detailed analysis of associated defects, or amniocentesis were not done, these issues do not seem to be the most important reason for the non-survivors.

In India, fetal echocardiography is not widely available yet. According to obstetrician's impressions, only 1-2% of pregnancies currently avail fetal echocardiography in less than 10 centers in the country. This proportion will surely increase. Recognizing the poor proportion of fetal survival, it is vital that the appropriate counseling be done by people well versed with the natural and unnatural history of the defects and knowledge regarding the associated abnormalities in the fetus as well, coupled with the grasp of the local realities. Even then, as shown in the study, the outcomes for fetuses may be suboptimal.

The decision regarding continuing pregnancy is difficult in the best of the circumstances in these situations. The role of counselors cannot be overemphasized. As is widely known, the information must be provided in a non-judgmental, yet sensitive manner. We must ensure that parents have understood the information and its implications fully and that any heart lesion is not equated with serious heart disease (and vice versa) before deciding.

The proportion of parents who elect to terminate pregnancy will depend on many factors including religion and cultural norms, as well as prognosis of cardiac lesion and associated non-cardiac anomalies. There is no agreement on the cardiac lesions for which termination may be advised because of several issues involved in the decision. In one of the European registry, termination was done in 6.6% after fetal cardiac diagnosis. It increased to 12-23% when a serious heart defect like hypoplastic left heart syndrome was diagnosed.[7] In the UK, between 1993 and 1995, half of the pregnancies affected by fetal cardiac defects resulted in termination.[8] In a study of 1006 fetuses who were diagnosed with congenital heart defects, survival rate after diagnosis was poor due to high termination rates and complexity of defects. Totally 558 (57%) parents chose to discontinue pregnancy. Chromosomal analysis was done in 412 fetuses out of which 42% proved to have anomalies.[9]

Early detection is usually associated with higher termination rate than the diagnosis made later in the gestation. In most of the European countries, the legal termination can be done up to 24 weeks of gestation. In India, the upper limit of legal termination is 20 weeks. But the medical termination of pregnancy (MTP) rules are quite liberal in India. In another Indian study from Chennai, out of 180 fetal echocardiography studies in a high-risk subset, congenital heart defects were found in 14 cases. Twelve fetuses had associated congenital abnormalities and all resulted in termination. [10]

Prenatal evaluation of the fetal heart has impacted practice of pediatric cardiology in several important ways. The incidence of serious congenital heart defects like hypoplastic left heart syndrome and pulmonary atresia with intact ventricular septum is declining. [4] This is attributed to high rate of fetal termination after fetal cardiac diagnosis. Increased rate of termination has raised concern in many western countries, especially regarding the future practice of pediatric cardiology. The analysis of fetuses with more severe defects like hypoplastic left or right heart syndrome in the patient population of the present study may be interesting, and may shed some more light on the dynamics of fetal survival.

Provision of adequate information about the problem

and results of available treatment is crucial in all cases. Most parents require continuing support and are likely to need more than one consultation in order to absorb and understand the implications of the ultrasound findings. Contact with other parents who have a child with similar problem may be helpful in decision-making. In a study by Bjorkhen, 64% of families who had lost a child due to heart defect previously were influenced in the decision of having another child because of availability of fetal echocardiography.^[11] Prenatal diagnosis is expected to make a positive impact on the clinical outcome of fetuses.^[12]

But the Indian realities are different. The liberal MTP rules, human resource crunch, financial difficulties, and lack of infrastructure all conspire against the chance of the fetus with heart disease to survive. It is even less likely for a female fetus although sex determination is legally prohibited. A right technology should have ensured better survival for the index case, but here is a paradox that is reflective of complexities of modern medicine in some ways. The study by Vaidyanathan *et al.* has done a stellar service by providing the data and focusing the attention on the fetus with heart defects.

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