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Fetal Cardiac Anomalies : A Fetal Echocardiographic Study

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= Abstract = **Seven years' experience of fetal echocardiography was reviewed to assess the diagnostic accuracy of fetal echocardiography, distribution of individual cardiac anomalies, and some practical problems of fetal echocardiography in Korea.**

A total of 737 fetuses were examined and 45 confirmed and 20 unconfirmed cardiac structural anomalies were identified. In general, fetal echocardiography was accurate in assessing fetal cardiac anomalies. However there were three false positive and four false negative diagnosis. In addition, diagnosis was changed slightly in 10 cases. Common errors occurred in the diagnosis of atrial isomerism, anomalous pulmonary venous drainage, complex atrioventricular junctional anomaly, ventricular septal defect, and coarctation of aorta. Some minor errors in diagnosis seemed unavoidable but this rarely had an important effect on prognosis. The common cardiac anomalies were ventricular septal defect, tetralogy of Fallot, and hypoplastic left heart syndrome.

Fetal echocardiography in Korea is at an early stage and does have many problems. Some of the problems are 1) insufficient number of doctors capable of doing fetal echocardiography, 2) few cardiac pathologists, 3) people's attitude toward congenital anomalies, 4) high rate of termination of pregnacy when fetal anomaly is found, and 5) doctors' lack of interest in confirming anomalies in stilborn fetuses or terminated products of pregnancy. Continuing education to the society and medical personnel seems urgent to solve some of these problems.

Key words : *fetus, cardiac anomaly, fetal echocardiography*

INTRODUCTION

Recent advances in two-dimensional and Doppler echocardiography permit accurate examination of the heart of the fetus (Kleinman et al. 1980; Allan et al. 1980; Allan et al 1984). With further improvement of technology, the capability for such evaluation is likely to increase in the future. The more we understand the nature and natural history of fetal cardiac anomalies, the better care we can provide to the mother and the fetus (Allan et al. 1986; Hornberger et al. 1991; Sharland et al. 1991). The pediatric cardiologist as well as obstetrician and radiologist are involved with this new population of patients and the cooperation of all involved is essential for total care of mother and fetus.

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The technique, accuracy, safety, and clinical importance of fetal echocardiography have been reported (Allan 1986; Oberhaensli et al. 1989; Wheller et al. 1990). The diagnosis of amajor cardiac defect in utero makes it possible for a mother to arrange delivery at a center with appropriate pediatric cardiovascular services. As an alternative, mother can make an informed decision concerning management of the pregnancy. In addition to identification of structural anomalies, accurate diagnosis of fetal cardiac arrhythmia can allow for successful transplacental treatment (Kleinman and Copel 1993). Fetal cardiac defects can occur in isolation, but they can coexist with other organ anomalies. Therefore identification of cardiac defects prompts thorough search of other organ anomalies and vice versa (Copel et al. 1986; Bert et al. 1988).

The incidence of cardiac anomalies as a whole is not different among various ethnic groups. However the incidences of individual anomalies may be different among different races (Ando and Takao 1978). Well known example is the higher incidence of subpulmonic ventricular septal defect in Asians. The incidence of individual cardiac anomalies in fetal population may be different from the incidence of postnatal population, because some cardiac anomalies incompatible with fetal survival might be less represented in postnatal studies (Allan et al. 1986). In Korea, a few centers are practicing fetal echocardiography and a systematic analysis of fetal cardiac anomalies has not been reported. This study was performed to review our 7 years' experience of fetal echocardiography and it was hoped that these initial results might help or encourage other centers in Korea to start the fetal echocardiographic program.

MATERIALS AND METHODS

We began fetal echocardiography in 1987 and 737 fetuses from 727 mothers have been studied so far. Two examinations per mother have been routine. All the pregnant mothers who underwent fetal echocardiography were asked to bring babies to our laboratory for postnatal confirmation or to inform us of cardiac status after delivery. However postnatal confirmation of cardiac status was not possible in some cases.

Study material consisted of all the consecutive fetuses identified to have cardiac anomalies by fetal echocardiography and all the products of pregnancy confirmed to have cardiac anomalies by postnatal echocardiography, cardiac catheterization, cardiac surgery, or autopsy. Fetal arrhythmias without cardiac anatomic defect were not included.

All the studies were performed with an Advanced Technology Laboratories Ultramark 8. Five mega Hz annular transducer was used for two-dimensional echocardiography and 3.0 or 5.0 M Hz mechanical transducer was used for a Doppler examination(Color Doppler was not available).

Fetal echocardiography was performed transabdominally with mothers supine. After identifying fetal position, a series of horizontal, saggittal and intermediate two-dimensional images were obtained. A systematic approach was used to make a cardiac diagnosis. In brief, situs of atrium, ventricle, and great artery were determined first and then individual component of every cardiac structure was assessed one by one. Doppler examination was performed when necessary to identify valvular regurgitation and abnormal flow direction.

RESULTS

Of 737 fetuses from 727 mothers, 64 fetuses were thought to have cardiac anomalies through fetal echocardiography. Of these 64, 44 fetuses had postnatal cardiac examinations and the rest 20 fetuses had no postnatal cardiac examinations. Among 44 fetuses who had postnatal confirmation, 41 were confirmed to have cardiac anomalies and three were proved to have normal heart (Fig. 1). Of those 673 fetuses who were thought to have normal heart, four were found to have cardiac anomalies through postnatal cardiac examinations. Therefore, of those 737 fetuses, 45 had confirmed cardiac anomalies and another 20 had unconfirmed cardiac anomalies. Although fetal echocardiography is not absolutely accurate, 20 fetuses with unconfirmed cardiac anomalies will also be des-

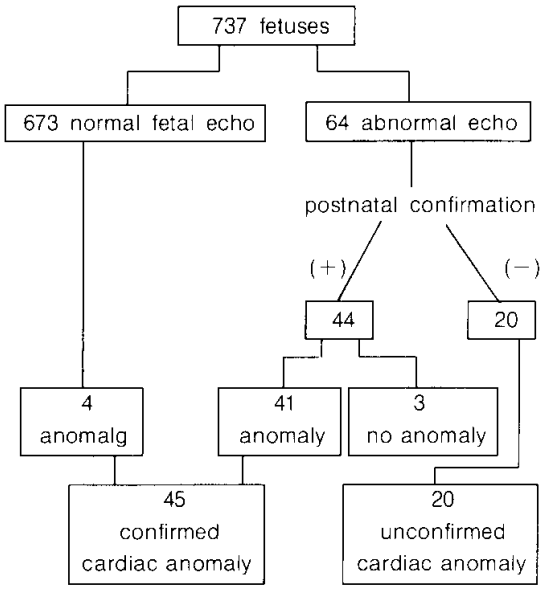

Fig. 1. Flow diagram of study population

Table 1. Maternal characteristics and indications of fetal echocardiography in fetuses with cardiac anomalies

	confirmed cases (45)	not confirmed cases (20)
Maternal age (year)		
median (range)	28 (21-36)	28 (23-37)
Gestation period (weeks)		
median (range)	28 (15-39)	27 (16-39)
Indication		
cardiac anomaly suspected by ob.	10 (22.2%)	6 (30.0%)
other organ anomaly	10 (22.2%)	5 (25.0%)
family history of CHD	9 (20.0%)	4 (20.0%)
fetal cardiac arrhythmia	8 (17.8%)	2 (10.0%)
fetal hydrops, ascites	6 (13.3%)	3 (15.0%)
drug ingestion and others	2 (4.4%)	0 (0.0%)

CHD; Congenital Heart Disease, ob.; Obstetricians

cribed separately.

The age of mothers who had fetuses with abnormal heart ranged from 21 to 36 years with a median of 28 years. Gestational period at the time of examination ranged from 15 weeks to 39 weeks with a median of 28 weeks. The common indications of fetal echocardiography were fetal cardiac anomaly suspected by obstetric scan, other associated anomalies, and family history of congenital heart disease (Table 1).

Accuracy of fetal echocardiography

The accuracy of fetal echocardiography was assessed by comparing fetal echocardiographic findings with postnatal findings. The differences between pre- and post-natal findings were found in 17 cases (Table 2). There were three false positive results. One fetus showed small aortic isthmus without other intracardiac abnormalities and coarctation was suggested. The baby was born in another hospital and did not show any symptoms or abnormal physical findings during the first 3 days after birth. Echocardiography was not done and further follow up examination was not performed. The other two false positive cases were thought to have small ventricular septal defect but image quality of both examinations was very poor because of later gestational period and unsatisfactory fetal position.

There were four false negative examinations (one case each of atrial septal defect, bicuspid aortic valve, ventricular septal defect, and coarctation of aorta). Retrospective review of video recordings in cases with atrial septal defect and bicuspid aortic valve did not show any diagnostic clue even with known diagnosis. Retrospective review of the case with ventricular septal defect revealed perimembrous ventricular septal defect which was overlooked during fetal echocardiographic examination. In the case with coarctation, the examination was given up, because it seemed impossible to visualize fetal heart due to omphalocele and improper fetal position.

In another 10 cases, the description of anatomic defects was not entirely correct. Among 3 cases with atrial isomerism, only one case was correctly diagnosed prenatally and two other cases were misdiagnosed as atrial solitus or inversus. The errors in the diagnosis of complex a-

Table 2. Differences between prenatal and postnatal cardiac diagnosis

Fetal echocardiography	Postnatal diagnosis
Small aortic isthmus (COA)	P/E was normal. echocardiography not done.
Suspicious small VSD	Normal
Suspicious small VSD	Normal
Normal fetal echo	ASD
Normal fetal echo	Bicuspid aortic valve
Normal fetal echo	VSD
Examination abandoned	COA
Atrial situs; inversus or ambiguous, Pulmonary venous drainage; uncertain	Left atrial isomerism, ipsilat. pul. ven. drain.
Atrial situs; solitus, Pulmonary venous drainage; uncertain	Left atrial isomerism, TAPVR to rt. sided at.
Aortic atresia with VSD	Aortic atresia with AVSD
DORV, AVSD, COA	DORV, mit. atr. with TV straddling, IAA
Aortic atresia, mitral atresia	Severe PS, normal MV
D-TGA	D-TGA with VSD
TOF	TOF with Cantrell's pentalogy
VSD	VSD+ASD
Severe aortic stenosis	Aortic atresia
Generalized aortic hypoplasia	COA

ASD; atrial septal defect, at.; atrium, atr.; atresia, AVSD; atrioventricular septal defect, COA; coarctation of aorta, D-TGA; complete transposition of the great arteries, DORV; double outlet right ventricle, drain.; drainage, IAA; interruption of aortic arch, ipsilat; ipsilateral, mit.; mitral, MV; mitral valve, P/E; physical examination, PS; pulmonic stenosis, pul.; pulmonary, rt.; right, TAPVR; total anomalous pulmonary venous return, TOF; tetralogy of Fallot, TV; tricuspid valve, ven.; venous, VSD; ventricular septal defect

trioventricular junctional anomalies occurred in 3 cases. Other errors were failure to recognize associated intracardiac defects such as ventricular septal defect or atrial septal defect.

Distribution of fetal cardiac anomalies

The distribution of cardiac anomalies is contained in Table 3. The total number of fetuses with cardiac anomalies was too small to draw conclusions but it was interesting to note that there were 5 confirmed cases of hypoplastic left heart syndrome, which is very rare in postnatal Korean series. Other common defects were ventricular septal defect and tetralogy of Fallot.

Family history of congenital heart diseases

There were 354 mothers who had a previous child or children with congenital heart disease. Among them, 12 mothers had recurrences of congenital heart diseases, which were confirmed in 8 and not confirmed in 4 cases. Overall recurrence rate was 3.4%. One mother who had a previous child with rhabdomyoma had a fetus with ventricular septal defect. Cardiac lesions in previous children and current fetuses were summarized in table 4. The cardiac lesions were concordant in 5 cases.

DISCUSSION

The diagnosis of fetal cardiac diseases has been possible with fetal echocardiography (Kleinman et al. 1980; Allan et al. 1980; Allan et al. 1984). Nonetheless there have been few thera-

Table 3. Distribution of cardiac anomalies

	confirmed cases	unconfirmed cases
Atrial isomerism	3 (6.7 %)	3 (15.0 %)
Atrial situs inversus	0 (0.0 %)	3 (15.0 %)
Single ventricle	1 (2.2 %)	1 (5.0 %)
HLHS (mitral or aortic atresia)	5 (11.1 %)	1 (5.0 %)
Tricuspid atresia	1 (2.2 %)	0 (0.0 %)
Truncus arteriosus	2 (4.4 %)	1 (5.0 %)
DORV	2 (4.4 %)	0 (0.0 %)
D-TGA	1 (2.2 %)	0 (0.0 %)
PA + intact ventricular septum	3 (6.7 %)	0 (0.0 %)
TOF	5 (11.1 %)	0 (0.0 %)
VSD + PA	0 (0.0 %)	1 (5.0 %)
AVSD	2 (4.4 %)	1 (5.0 %)
COA	3 (6.7 %)	2 (10.0 %)
VSD	5 (11.1 %)	5 (25.0 %)
VSD + absent pulmonary valve	0 (0.0 %)	1 (5.0 %)
ASD	1 (2.2 %)	0 (0.0 %)
Other developmental anomalies	3 (6.7 %)	1 (5.0 %)
Cardiomegaly + TR	5 (11.1 %)	0 (0.0 %)
Rhabdomyoma	3 (6.7 %)	0 (0.0 %)
Total	45 (99.9 %)	20 (100.0 %)

HLHS; hypoplastic left heart syndrome, PA; pulmonary atresia, TR; tricuspid regurgitation. The other abbreviations are the same as in table 2.

Table 4. Cardiac lesions in cases with positive family history cardiac lesions in pre. children confirmed lesions in current exam.

Cardiac lesions in pre. children	Confirmed lesions in current exam.
D-TGA	ASD
L-TGA	D-TGA
Rhabdomyoma	VSD
VSD	TOF
Two children; both unspecified	PA + intact ventricular septum
VSD	VSD
Tricuspid atresia	Tricuspid atresia
Truncus arteriosus	Truncus arteriosus
AVSD	AVSD
Cardiac lesions in pre. children	**Unconfirmed lesions in current exam.**
TOF	VSD
Two children; both truncus	VSD + PA
D-TGA,AVSD, pul.atr.	mirror image dextrocardia
VSD	VSD

exam.; examination, pre.; previous, L-TGA; corrected transpositions of the great arteries

peutic trials to treat fetal cardiac diseases (Maxwell et al. 1991). One of the most important implications of fetal echocardiography may be prognostic stratification of fetal cardiac diseases. Since there is no other way but fetal echocardiography to diagnose fetal cardiac diseases and all the decisions should be made on the basis of fetal echocardiographic findings, it is very important to realize the limitation of fetal echocardiography in terms of diagnostic accuracy. In addition, it is also very important to realize that our knowledge on the pathophysiology and the natural history of fetal cardiac diseases is not sufficient and we are beginning to gain some insight into pathophysiology and natural history of fetal cardiac diseases. Therefore extreme care should be exercised when we council the parents.

Although there have been many reports of fetal echocardiography, a few systematic studies were performed to examine the diagnostic accuracy of fetal echocardiography (Benacerraf et al. 1987; Allan et al. 1989). The diagnostic accuracy of postnatal echocardiography in children has been documented in many studies (Gutgesell et al. 1985; Krabill et al. 1987) and it is generally accepted that postnatal echocardiography can provide images of anatomic structures in such detail that cardiac surgery can be performed without cardiac catheterization (Leung et al. 1986; Sreeram et al. 1990). Fetal echocardiography can not be compared to postnatal echocardiography in terms of accuracy, because fetal heart is far away from the transducers, the the fetal parts or umbilical cords are often interfering with cardiac imaging, and orientation of images are not always clear.

The echocardiographic determination of atrial situs is by inferrence from the relative position of abdominal aorta and inferior vena cava. This is easy in postnatal echocardiography. However it is not easy in some fetal echocardiography and ambiguous atrial situs may be misdiagnosed as solitus or inversus. The determination of ventricular situs is not difficult as long as there are two identifiable ventricles. Unlike a postnatal heart, the pulmonary artery in a fetus also forms an arch with ductus arteriosus and descending aorta and there are two vascular arches, one aortic arch and the other ductal arch. Nonetheless the identification of the great arteries through fetal echocardiography is not difficult when there are two patent arteries. The direction of the proximal portion of the great arteries, shape and height of arch, and most importantly the branching pattern of the great arteries are all helpful to identify which is which. However when a single great artery arises from the heart, it may be difficult to determine whether the single artery is an aorta, a pulmonary artery or a truncus arteriosus. Careful identification of branches may solve the problem.

In fetal life, foramen ovale in atrial septum is widely patent and it is often difficult to decide whether there is an atrial septal defect. Total anomalous pulmonary venous return can occur in isolation or as a component of complex cardiac anomalies. When it occurs in isolation, it could be diagnosed through fetal cardiac scan but if it is a component of complex cardiac anomalies, it is often very difficult to make a diagnosis of it. Atrioventricular junctional morphology is easily recognizable and a four chamber view may be the easiest view to obtain. However when there are unusual anomalies such as unequal division of atrioventricular junction with or without atrioventricular septal defect or one valve atresia, there is a some chance of misinterpretation. Mitral atresia or tricuspid atresia might be mislabeled as atrioventricular septal defect or vice versa. When only one ventricle is identified, it may be difficult to exclude the presence of other very hypoplastic ventricle and a diligent search for a possible small ventricle should be done. Ventricular septal defect can be identified when the defect is large but may not be found if the defect is small or in unusual position. Doppler or color Doppler may or may not be helpful in identifying small ventricular septal defect.

Application of Doppler and color Doppler to cardiovascular system makes it possible to get hemodynamic information. Major application of Doppler in fetal echocardiography is the detection of abnormal flow direction such as atrioventricular regurgitation, ductal left to right shunt or

atrial left to right shunt (Sharland et al. 1990; Copel et al. 1991). Doppler assessment of valvular stenosis is very useful in postnatal heart but because of parallel circulation of the fetal heart, Doppler examination of fetal stenotic valve does not usually show higher velocity. Severe aortic stenosis usually manifests as a dilated poorly contracting left ventricle and it is difficult to differentiate severe aortic stenosis from primary left ventricular dysfunction (Sharland et al. 1991). Mild or moderate semilunar valve stenosis is difficult to diagnose because it is usual not to have higher velocity by Doppler technique.

In summary, the echocardiograpahic diagnosis of structural heart disease can be achieved with accuracy in fetal life. It is likely, nonetheless, that the prenatal echocardiogram will continue to be less accurate than postnatal echocardiography, and both pulsed and color Doppler are not as useful tools in terms of improving accuracy as they are after birth.ms of improving accuracy as they are after birth. Image quality at the study, which may be reduced by early or late gestation or maternal obesity, will influence precision. Some minor errors in diagnosis may be unavoidable but this will rarely have an important effect on prognosis. Diagnosis can be sufficiently precise for management decisions to be based on the echocardiogram prenatally as is now the practice in postnatal life (Allan et al. 1989).

While fetal echocardiography has been being practiced for more than 15 years in advanced countries, there are a few centers in Korea practicing fetal echocardiography. As cardiac anomaly is one of the most frequent fetal anomalies and determines the prognosis of the fetus, more fetal echocardiography should be performed in Korea. In order to achieve that purpose, more obstetricians and radiologists should be motivated, educated and trained in established centers. Another problem in Korea is that there are few cardiac patholgists interested in congenital cardiac anomalies. Since congenital cardiac defects are very varied and moreover the fetal heart is not only small but also may show very unusual anatomic defects, fetal cardiac specimens should be throughly examined by interested cardiac pathologists. Otherwise incomplete diagnosis may result even after pathologic examinations.

Significant proportion of fetuses with abnormal fetal echocardiographic findings in our study population was not confirmed after pregnancy was ended. We ask every mother to bring their babies to our laboratory or to inform us of cardiac conditions after delivery. When the fetuses are expected to be born, we try as hard as we can to get follow up information of the mothers and the fetuses. In spite of all these efforts, we could not get post natal information in 20 mothers who had fetuses with abnormal fetal echocardiography. The reasons for failure to get follow up information were manyfold. The most important reason may be people's attitude toward congenital anomalies. They usually do not want to have babies with congenital anomalies and they do not seem to realize the importance of confirmation of the diagnosis and of providing the information. As a result, when they are told that their pregnancies are complicated by fetal anomalies, they just want to terminate the pregnancy, which is possible at many clinics throughout Korea though it is illegal after 25 weeks of gestational period. Another reason may be a lack of interest among doctors. Autopsy rate of still birth or terminated product is very low. Continuing education to the public and medical personnel seems necessasy to improve this situation.

Prenatal diagnosis of genetic diseases and organ anomalies will expand very rapidly (Desforges 1993) and it will inevitably create many ethical problems (Veille et al. 1989). In order to improve maternal as well as fetal well-being in complicated pregnancies, all the involved medical branches should cooperate. Likewise, joint effort of society and the medical field seems essential to solve many ethical problems arising from prenatal diagnosis.

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