Identification of congenital cardiac malformations by echocardiography in midtrimester fetus*

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SUMMARY Fetal echocardiograms were performed in 21 pregnancies before midtrimester termination. All fetal heart specimens were collected and studied morphologically. Eighteen had been diagnosed as normal echocardiographically and this was confirmed anatomically.

Deficiency of the atrial septum was suspected in one, and primum and secundum atrial septal defects were found anatomically. Coarctation of the aorta was suspected in another and this was confirmed anatomically. A ventricular septal defect was suspected in one which proved to be normal on dissection.

The anatomical features of the normal heart in the midtrimester fetus can be identified using two-dimensional echocardiography.¹ The absence of air-filled lungs makes the fetal heart more accessible to ultrasonic examination than is the case in postnatal life. It is thus possible to visualise the heart and great vessels "in the round". This makes it easier to trace the course and the connections of the great vessels to and from the heart and to determine the connections of the chambers within it. It follows that it should be possible to diagnose abnormalities of cardiac connections and, given the resolution of the technique, abnormalities of cardiac septation too. To evaluate further the accuracy of the technique, we studied, echocardiographically, 21 fetuses before midtrimester termination and then compared the echocardiographic with the anatomical findings.

Patients and methods

A group of 21 patients who were about to undergo midtrimester termination of pregnancy were studied echocardiographically using an ATL Mark III sector scanner or Kretz Combison 100 sector scanner. The pictures were recorded on a Sanyo VTC 7100 video cassette recorder and subsequently analysed. The indications for termination of pregnancy are given in the Table. The anatomical specimens were obtained from one day to three weeks after the echocardiographic examination and were dissected to provide direct comparison with the echocardiographic find-

Table

Reason for termination	Number
Social	8
Neural tube anomalies	4
Other non-cardiac congenital abnormalities	3
Down's syndrome	4
Turner's syndrome	1
Rh isoimmunisation (intrauterine death)	1

ings. The gestational ages ranged from 16 to 27 weeks.

Results

After echocardiographic examination, 18 fetal hearts were considered to have two atria and two ventricles normally connected, together with normal venous and arterial connections. The septa were considered to be intact apart from the mechanism of the foramen ovale. Three fetal hearts were considered to be abnormal at echocardiographic examination. The moving real time images are much easier to interpret than the static pictures. This is especially so before 24 weeks gestation when the fetal heart is very small. One fetus, of 18 weeks gestation, was thought to have an inlet septal ventricular defect echocardiographically (Fig. 1), but was found to be anatomically normal. Even retrospective study of the echocardiographic pictures after the anatomical dissection did not absolutely exclude a ventricular septal defect.

Echocardiograms from the second heart thought abnormal and studied at 16 weeks gestation showed

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Fig. 1 Four chamber projection of 18 week gestation fetal heart. There appears to be a defect at the top of the ventricular septum.

deficiency of the atrial septum, and the two atrioventricular valves were inserted at the same point in the ventricular septum (Fig. 2a and b). The difference in septal insertion of the mitral and tricuspid valves on a four chamber projection of the fetal heart is an important normal finding. It can be appreciated in most cases by the difference in angulation of the two atrioventricular valves, particularly seen in the moving images. In our illustrated examples, in the atrioventricular defect the two atrioventricular valves lie straight across the septum (Fig. 2a). In contrast, in the normal heart the differential insertion of the two valves is represented by a "skewed" orientation (Fig. 3, top). In the apical four

Fig. 3 (top) Four chamber projection of the fetal heart illustrating the differential septal insertion of atrioventricular valves in the normal fetal heart. (bottom) Corresponding anatomical section of a normal fetal heart.

Fig. 2(a) Echocardiographic four chamber projection of 16 week fetal heart showing the presence of an atrioventricular defect. (b) Corresponding anatomical section of the same fetal heart.

chamber view seen in Fig. 2a the atrial septum often shows dropout but on no projection of the heart with the atrioventricular defect could any atrial defect be detected. The lateral view of the ventricular and atrial septum in the fetus, which corresponds to the subxiphoid view in postnatal life, is more reliable for the complete visualisation of both septa. This fetus had Down's syndrome diagnosed by chromosomal analysis of an amniotic fluid specimen. Anatomical dissection disclosed an atrioventricular defect with separate valve orifices (ostium primum atrial septal defect) together with a secundum atrial septal defect (Fig. 4). There was only a narrow strip of atrial septum between the ostium primum defect and the secundum defect, and, as expected in an atrioventricular defect, the right and left valves were attached to the septum at the same level (Fig. 2a and b).

Echocardiograms from the third fetal heart studied at 18 weeks gestation showed that the right ventricle was larger than the left. The left ventricle was judged to be smaller than expected for the gestational size, though this judgement was difficult in view of the right-sided dilatation (Fig. 5). The fetus had ascites and oedema, presumably secondary to cardiac failure. An M-mode echocardiogram across the two ventricular chambers showed that the right ventricular cavity was larger than the left but there was an apparently normal atrioventricular valve in each chamber (Fig. 6). The ascending aorta was identified together with the descending aorta, ductus arteriosus, and the junction of the subclavian artery

Fig. 4 Anatomical dissection of the heart illustrated in Fig. 2. There is an ostium primum atrioventricular defect together with a secundum atrial septal defect. The heart is opened from the right side.

with the descending aorta (Fig. 7). In spite of a careful search the arch of the aorta was not seen. Though the full extent of the aortic arch is not seen on every examination it can be seen in every normal patient when searched for diligently. This is particularly so during the optimum time for fetal echocardiography, that is at between 20 and 26 weeks gestation. When the ascending aorta, the subclavian artery, and the descending aorta have all been seen in the same echocardiographic section, the arch has always previously been identified. Therefore in this par-

Fig. 5 Two-dimensional echocardiogram in four chamber projection demonstrating dilatation of the right atrium and right ventricle.

Fig. 6 *M-mode echocardiogram from the same fetus as shown in Fig. 5 taken across two ventricular chambers showing dilatation of the right side of the heart.*

ticular case the absence of a segment of arch raised a high index of suspicion for the diagnosis of isthmal hypoplasia. The fetus was found on chromosomal analysis to have Turner's syndrome. Dissection of the fetal heart disclosed gross hypoplasia of the aortic arch between the left common carotid and the left subclavian arteries (Fig. 8). Dilatation of the right side of the heart was confirmed, the left ventricle being more or less normal in size.

Fig. 8 *The anatomical specimen from the 18 week fetus shown in Fig. 7 illustrating gross tubular hypoplasia of the aortic arch between the left common carotid and the left subclavian artery.*

Discussion

If fetal echocardiography is to play a role in the management of congenital cardiac malformations, it is essential to determine the accuracy of the technique. This requires certainty in the diagnosis of normal hearts as well as hearts containing congenital lesions. We are aware that cases of hypoplastic right heart and univentricular heart have been diagnosed in fetuses of

Fig. 7 *Echocardiogram showing the descending aorta in an 18 week fetus with hypoplasia of aortic arch.*

34 to 38 weeks gestation,² while single cases have been identified in the middle trimester with a uni-ventricular heart³ and pulmonary atresia with ventricular septal defect (J Roelandt, 1980, personal communication) respectively. All the fetal echocardiograms were performed once only and 15 out of 21 fetal hearts were less than 20 weeks gestation. Though there is usually a time limitation on a particular examination, there is normally an opportunity for restudy slightly later. Therefore any structure not identified on the first study, for example, the aortic arch, can be searched for from the start of the next examination. In this study we examined the hearts of fetuses which were at particularly high risk of having cardiac malformations. We successfully diagnosed normality in 18 hearts and identified correctly cardiac lesions in two hearts with normal cardiac connections. We were unsuccessful in only one case, diagnosing an inlet ventricular septal defect when the septum was intact. It is significant that the two lesions diagnosed might not be considered "obvious", and that the pregnancies were examined at 16 and 18 weeks gestation. Thus, in the first case an atrioventricular defect with separate valve orifices (ostium primum atrial septal defect) was successfully identified while in the second case it proved possible to predict either tubular hypoplasia or interruption of the transverse portion of the aortic arch. These findings suggest that more gross malformations, such as abnormal atrioventricular connections or common valves etc., should be detectable. We also suggest on the basis of this experience that abnormalities of cardiac septation should be diagnosed. In one case, however, we did diagnose a ventricular septal defect when none existed. Even on retrospective examination of the videotape from this pregnancy it was not possible to be sure that the septum was intact (Fig. 1). The presumed defect had been identified in the four-chamber plane viewed from the cardiac apex. We now

know that the same four chamber plane can be obtained with the echo beam at right angles to the inlet septum, producing an appearance similar to the subxiphoid four chamber view used in paediatric practice. In future we will not diagnose such a ventricular septal defect unless it is visualised in both the "apical" and "lateral" views of the septum.

It remains to be seen if it is possible to diagnose abnormalities of ventriculoarterial connections such as complete transposition. To do this it will be necessary to distinguish unequivocally a morphologically right ventricle from a morphologically left ventricle and to trace both ventricles to the great arteries they support. We think this is possible, but we are now engaged in assessing the criteria for ventricular and great arterial identification in further pregnancies.

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