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Atresia or absence of the left-sided atrioventricular connection in the fetus: echocardiographic diagnosis and outcome

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ABSTRACT

Our objective was to describe the appearance of atresia of the left-sided atrioventricular valve on the four-chamber view of the fetal heart. Twenty-four cases of atresia of the left-sided atrioventricular connection were detected over a 2-year period. Cases were analyzed according to the appearance of the four-chamber view and the connections of the great arteries.

All cases were referred because the left-sided ventricle was smaller than the right. There were five different groups of cardiac anomaly detected. Associated findings varied according to the diagnostic group.

It was concluded that it is possible to identify atresia of the left-sided atrioventricular connection accurately during obstetric ultrasound examination by evaluation of the four-chamber view. However, the appearance of this view varies with the diagnosis. Each diagnostic group has different implications in terms of associated extracardiac malformations and postnatal management.

INTRODUCTION

It has become well established that congenital heart disease can be detected accurately in the fetus from as early as 14 weeks' gestation[1–3]. Since evaluation of the four-chamber view during obstetric scanning was first promoted in the mid-1980s, cardiac malformations have increasingly been detected by the sonographer and referred to the pediatric cardiologist for more complete diagnosis[4,5]. Cardiac malformations, nonetheless, remain the most common lesions overlooked during routine prenatal ultrasound examinations[6,7]. In the RADIUS study[8], no cardiac malformations were detected prior to 24 weeks' gestation outside the tertiary centers. Some authors have suggested that the great arteries should also be assessed during obstetric scanning; this would certainly improve the detection rate of cardiac malformations[9–11]. However, many cardiac abnormalities continue to be overlooked, despite abnormality in the four-chamber view.

We therefore review the normal features to be noted in the four-chamber view, and describe a group of cardiac anomalies with atresia or absence of the left-sided atrioventricular connection as the common feature. The variation in ultrasound findings from normal, and between groups within this heading, are illustrated.

MATERIALS AND METHODS

Over 300 patients were referred for fetal echocardiography between August 1993 and the end of 1995. There were 80 cases of fetal congenital heart disease detected, of which 24 were diagnosed as having an absent left-sided atrioventricular connection. All had an abnormal four-chamber view and were referred from the obstetrician or sonographer for this reason. The gestational age range was 16 to 37 weeks with a mean of 26.3. Twelve of the 24 were diagnosed prior to 24 weeks' gestation. The associated cardiac and extracardiac lesions were noted and the outcome of the pregnancy determined. Retrospective measurements were carried out of the width of the ventricular chambers just below the atrioventricular junctions in the anteroposterior diameter, the length of the ventricles from the atrioventricular junction to the apex and the width of the two great arteries at their origin. The ratios of the widths of the left ventricle to the right ventricle (LV/RV) and of the aorta to the pulmonary artery (Ao/PA) were compared to normal[12].

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RESULTS

The fetal cardiac diagnosis was confirmed echocardiographically in all 19 babies who were born alive. The prenatal diagnosis was the same as that made postnatally in all cases, although one case was redefined (from Group 2a to Group 2b) during fetal follow-up. Karyotype was available in 21 of 24 cases. Autopsy was not performed in four cases in which the pregnancy was interrupted and one in which there was a spontaneous intrauterine death. The results are summarized in Table 1.

The anatomical specimens used in the illustrations were drawn from our fetal collection and were chosen to match the gestational age and appearance of the echocardiogram. They do not represent the same cases as are illustrated in the ultrasound images.

Group 1: the hypoplastic left heart syndrome

There were 14 cases encountered with concordant segmental connections, mitral and aortic atresia, reversed interatrial shunt, hypoplasia of the left ventricle and aorta, and reversal of flow in the aortic arch. In four of the cases, there was a trivial amount of flow through the mitral valve in early pregnancy, but this had ceased by term, so these cases are included in this group. The four-chamber view in a typical example is illustrated in Figure 1.

Extracardiac anomalies These were found in three of 14 cases (21%), including one case of trisomy 18.

Outcome There were one termination of pregnancy and seven neonatal deaths. There was an 'intention to treat' in five of these seven. Six neonates survived the first-stage Norwood procedure, but one suffered severe neurological damage, related to surgery at another center, and will not

Table 1 Results of study

Case number	*Gestational age at diagnosis* (weeks)	*Karyotype*	*Outcome*
Group 1	*Hypoplastic left heart syndrome*		
1	23	normal	alive
2	31	normal	NND
3	36	normal	alive
4	21	N/A	TOP
5	34	normal	InfD (ECAs)
6	35	normal	NND
7	22	normal	NND
8	17	normal	NND (ECAs)
9	19	normal	alive
10	37	normal	alive
11	17	normal	alive
12	30	trisomy 18	NND
13	23	normal	NND
14	35	normal	alive
Group 2	*Concordant AV connections with DORV*		
Group 2a	*Mitral atresia, DORV*		
1	16	N/A	TOP
2	22	N/A	IUD
3	20	trisomy 18	TOP
Group 2b	*Common valve to RV, DORV*		
1	19	normal	InfD (ECAs)
2	30	normal	TOP (ECAs)
3	30	normal	InfD (ECAs)
Group 3	*Mitral atresia, VSD*		
1	31	normal	NND
2	2	normal	alive
Group 4	*Absent left, discordant univentricular heart*		
1	33	normal	alive
2	20	normal	alive

NND, neonatal death; N/A, not available; TOP, termination of pregnancy; InfD, infant death; ECAs, extracardiac anomalies; AV, atrioventricular; DORV, double-outlet right ventricle; IUD, intrauterine death; RV, right ventricle; VSD, ventricular septal defect

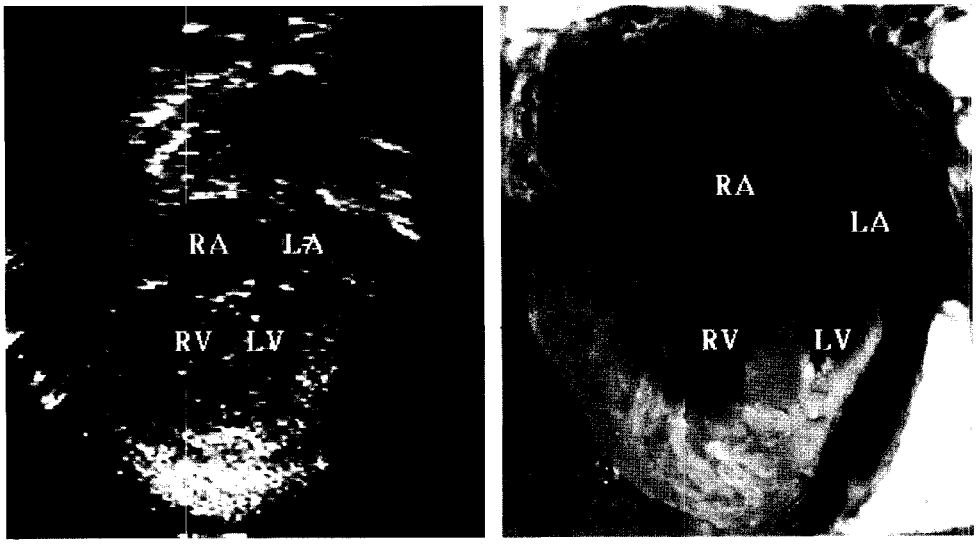

Figure 1 The fetal heart is seen in the four-chamber view echocardiographically with a matching anatomical specimen alongside. The left atrium is small and the left ventricular cavity diminutive. The mitral valve is hypoplastic, with no flow across it. Note that the right ventricle forms the apex of the heart. These findings are typical of the hypoplastic left heart syndrome. RA, right atrium; RV, right ventricle; LA, left atrium; LV, left ventricle

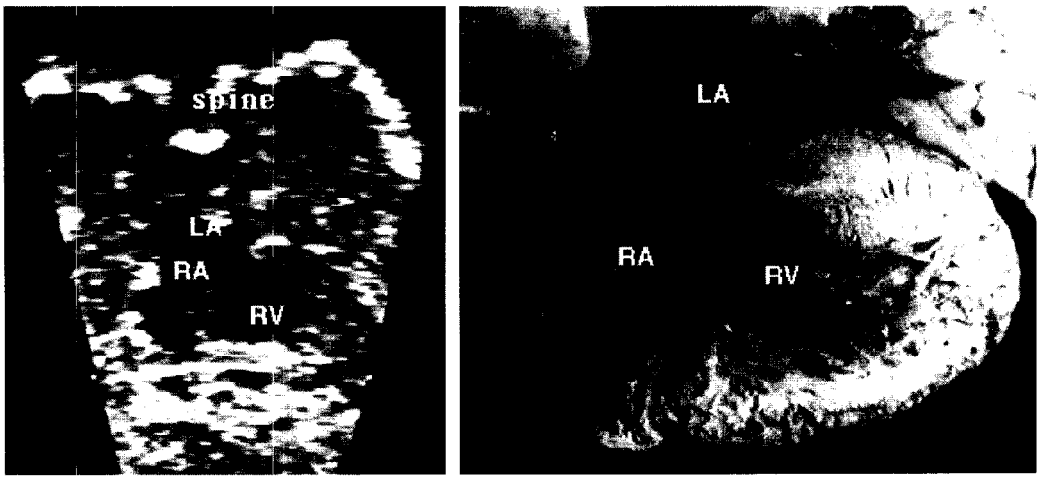

Figure 2 The fetal heart is seen in the four-chamber view echocardiographically with a matching anatomical specimen alongside. The left atrium (LA) is small. There is no mitral valve or left ventricular cavity distinguishable. This is a case of mitral atresia with a double-outlet arterial connection. RA, right atrium; RV, right ventricle

proceed to further surgery. Two of the deaths, one prior to and one after Norwood surgery, were caused by extracardiac lesions, tracheoesophageal fistula (one case) and multiple bowel atresias (one case).

Group 2: concordant atrioventricular connections with double-outlet right ventricle

Group 2a: with separate atrioventricular junctions

There were three cases of mitral atresia with double-outlet right ventricle. In all three, the left ventricle was diminutive or undetectable (Figure 2). The great arteries arose in parallel orientation with the aorta anterior and to the right of the pulmonary artery. There was valvar pulmonary stenosis in two of the three.

Extracardiac anomalies One of the three had multiple extracardiac malformations and trisomy 18; one further case was thought to have other anomalies, but the mother refused further evaluation; the fetus died *in utero* and autopsy was not performed.

Outcome Two mothers elected termination of pregnancy. There was one spontaneous intrauterine death. There are therefore no survivors in this group.

Group 2b: with common atrioventricular junction

There were three cases with the right component of a common atrioventricular valve connecting to a dominant right ventricle and an atretic or severely obstructed left-sided component. Particularly in early pregnancy, this was indistinguishable from cases in Group 2a in the four-chamber view. There was double-outlet right ventricle in all three cases, with the aorta anterior and to the right of the pulmonary artery. There was pulmonary stenosis in all three cases.

Extracardiac anomalies There was isomerism of the right atrial appendages in all three cases, with associated asplenia, a central liver and malposition of the intestines[13]. There were no chromosomal anomalies in this group.

Outcome One mother elected termination of pregnancy. One infant underwent placement of a Blalock shunt. Due to poor right ventricular function, the infant did not proceed to a Fontan procedure but was rerouted to transplantation. She died after this procedure. The remaining infant died after shunt placement.

Group 3: mitral atresia with ventricular septal defect

There were two cases of mitral atresia with a ventricular septal defect and concordant ventriculoarterial connection to a patent aorta. The great arteries were normally related. The mitral valve in both cases was imperforate, but the width of the left ventricle was only slightly smaller than the right and of normal length in one case (Figure 3). In this case, the left ventricle was supplied through a large muscular ventricular septal defect.

Extracardiac anomalies There were no extracardiac defects in these two cases.

Outcome Both neonates underwent the first-stage Norwood procedure, with one surviving.

Group 4: dominant left ventricle with absent left atrioventricular connection

There were two cases with atresia of the left-sided atrioventricular connection between the left atrium and

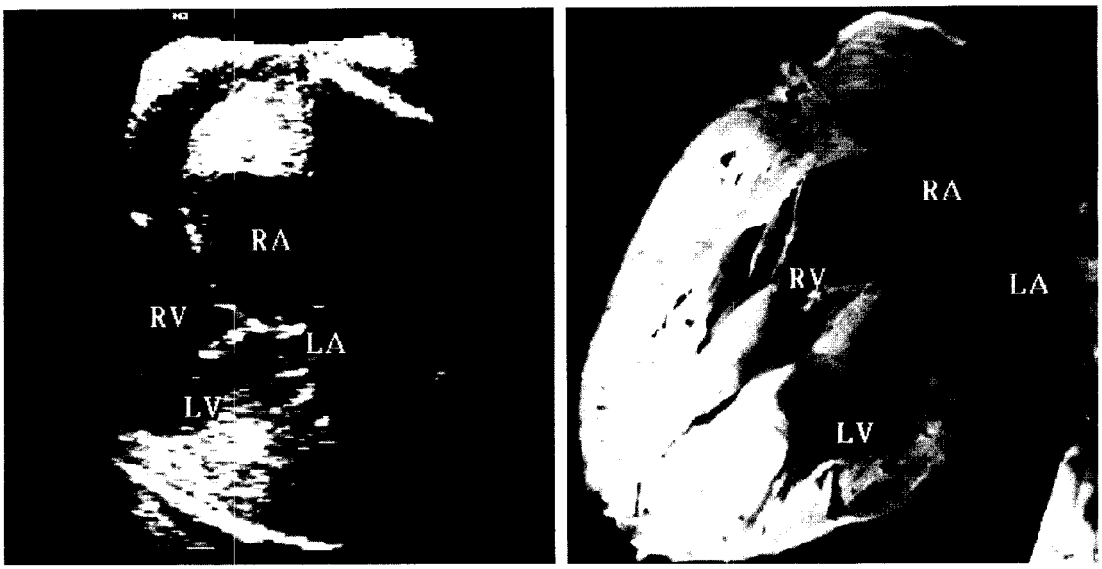

Figure 3 The fetal heart is seen in the four-chamber view echocardiographically with a matching anatomical specimen alongside. The left atrium (LA) and left ventricle (LV) are small. The mitral valve is imperforate. There was no flow through it to the cavity of the left ventricle. The left ventricle received blood only through the ventricular septal defect. RA, right atrium; RV, right ventricle

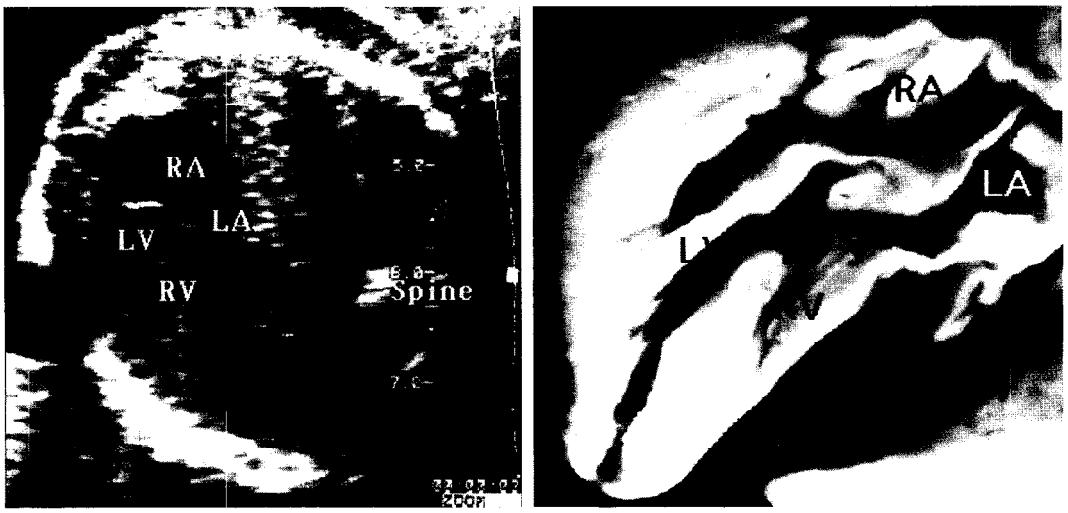

Figure 4 The fetal heart is seen in the four-chamber view echocardiographically with a matching anatomical specimen alongside. The right atrium (RA) is connected to a morphological left ventricle (LV) through a mitral valve. The left atrium (LA) is separated from the morphological right ventricle (RV) by an atretic left-sided connection. The rudimentary right ventricle is supplied only through the ventricular septal defect

left-sided ventricle, but the right atrium was connected to a dominant, morphological left ventricle through a patent right-sided atrioventricular valve (Figure 4). The pulmonary artery arose from the dominant right-sided left ventricle and the aorta from the rudimentary and incomplete right ventricle, which lay on the left lateral upper border of the heart. These cases, therefore, had a univentricular atrioventricular and discordant ventriculoarterial connections with absence of the left-sided atrioventricular connection. The great arteries arose in parallel orientation with the aorta to the left of the pulmonary artery. There was valvar pulmonary stenosis in one of the two.

Extracardiac anomalies There were no extracardiac defects in these two cases.

Outcome Both infants survived and one has undergone banding of the pulmonary trunk.

Measurement data

One case in Group 2 was unsuitable for measurement for technical reasons. The size of the left-sided ventricle in all other cases was assessed at the initial study and plotted in Figure 5. It can be seen that the width of the left-sided ventricle was markedly smaller than the right in all the diagnostic categories. In four cases, the left ventricle could not be measured, as no cavity could be detected. With the exception of one case in Group 4 already mentioned, the left-sided ventricle in all cases was less than one-half to one-third of the length of the right.

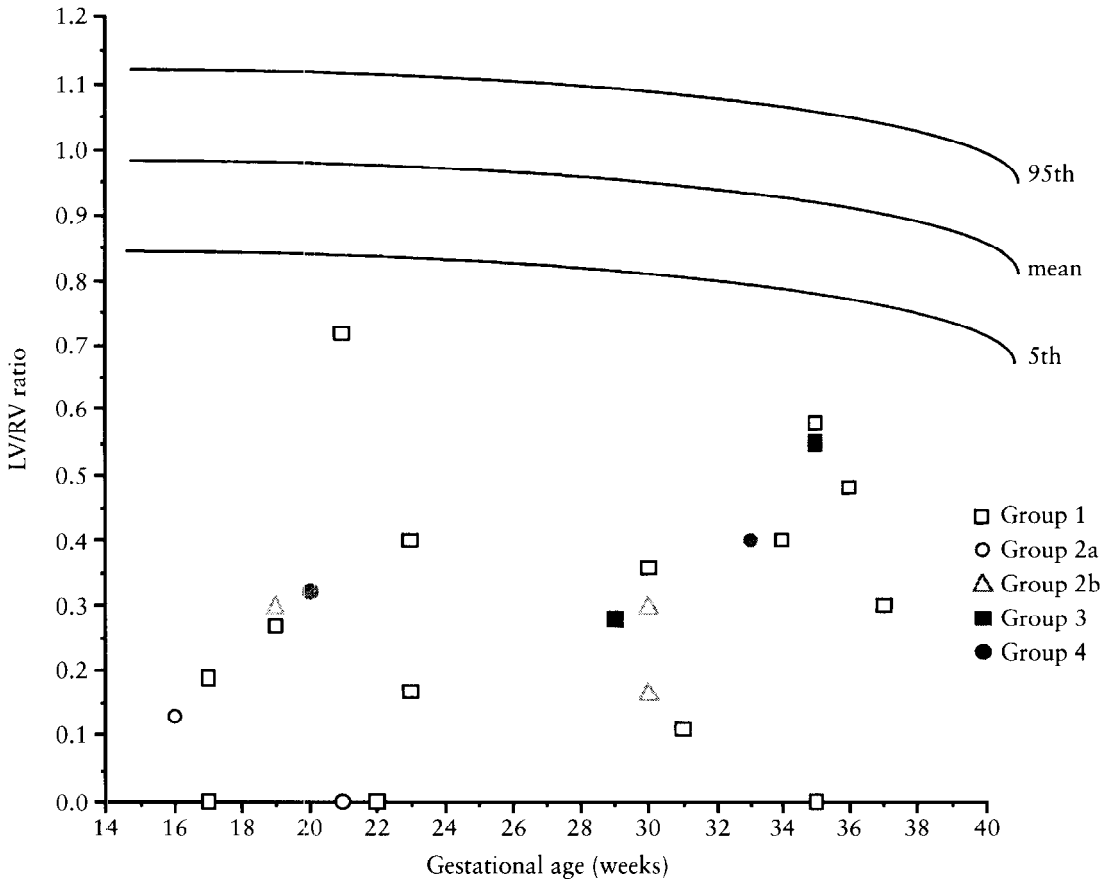

Figure 5 The ratios of the left ventricular to right ventricular width (LV/RV), measured just below the atrioventricular junctions, are plotted for each diagnostic group. The mean, 5th and 95th centiles of the LV/RV ratio in the normal fetus are shown for comparison. Although the left ventricle was consistently smaller than the right, the degree of hypoplasia varied

The width of the ascending aorta at the initial study relative to the pulmonary artery is shown in Figure 6. It can be seen that the aorta was dilated in one of each of the cases in Groups 2a and 4 and normal in size in the remaining cases in these two groups. In all cases in Group 2b, there was a high Ao/PA ratio. In the case in Group 4 with a large ventricular septal defect, the aorta was close to a normal size. In all other cases the aorta was significantly below the 5th centile.

DISCUSSION

The size of the ventricles and great arteries is mainly determined by the flow they receive. The size of the left-sided ventricle reflects either the time in gestation when the atrioventricular valve became critically obstructed and the left ventricle stopped growing normally as a result[14,15], or the size of a ventricular septal defect, if present. Similarly, the size of the aorta will be normal if there is unobstructed flow within it, as in a double-outlet connection or if a communicating ventricular septal defect is unrestrictive.

During an obstetric scan the evaluation of a normal four-chamber view should include assessment of:

(1) *Size*: the heart should occupy about one-third of the area of the thorax;

(2) *Position*: the apex should point out of the left anterior thorax with an angle of about 40° to the midline of the thorax;

(3) *Structure*: there should be two atria of equal size, two ventricles of equal size and thickness, and an intact crux; and

(4) *Function*: there should be two opening atrioventricular valves and two equally contracting ventricles[16,17].

Clearly, the four-chamber views in all 24 cases were abnormal by virtue of a smaller left-sided than right-sided ventricle, and no patent connection between the left atrium and left-sided ventricle. However, the degree of 'smallness' in width varied somewhat, as can be seen in Figure 5. With one exception, the length of the ventricle was abnormal in all cases. In addition, it can be seen from Figures 1–4 that the left atrium was also small. In coarctation of the aorta, or in unobstructed anomalous pulmonary venous connection, the left ventricle will also be smaller than normal in width, but of a normal length, and the LV/RV ratio is decreased[18]. However, the mitral valve is patent, differentiating between either of these diagnoses and an absent left connection.

The diagnostic categories were distinguished by their arterial connections. The great arteries were normally related, that is the pulmonary artery 'crossed over' the

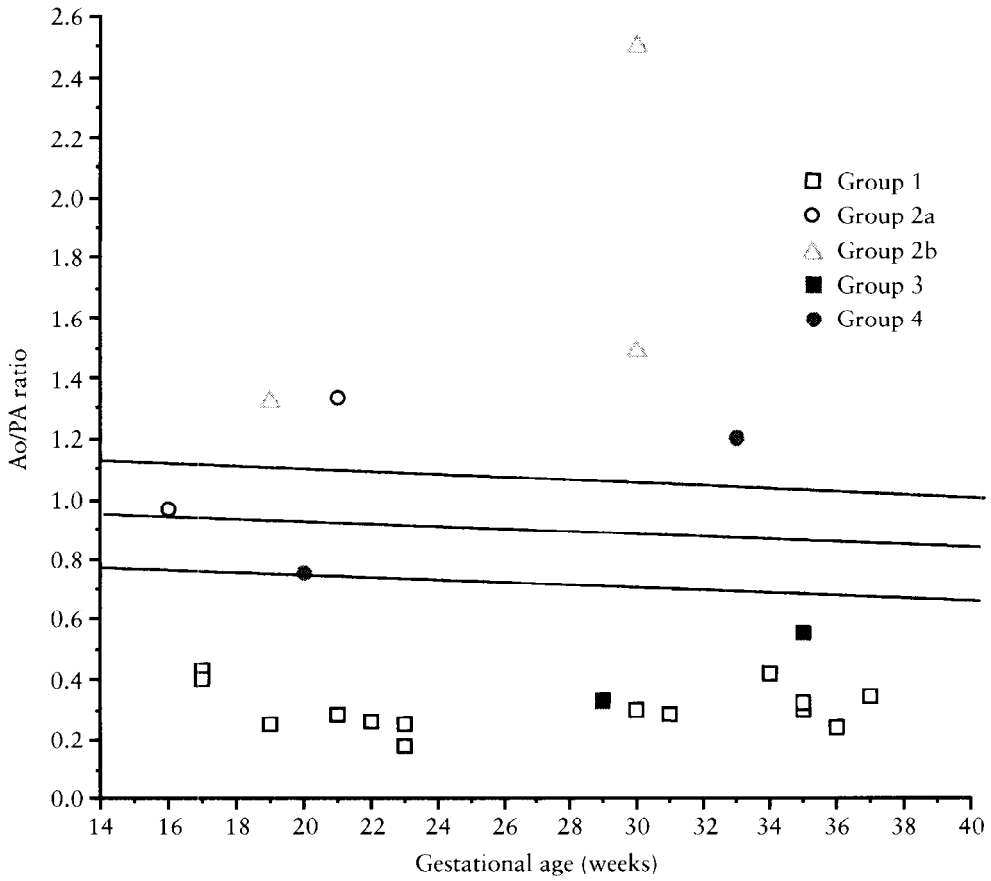

Figure 6 The ratio of the width of the aorta (Ao) and the pulmonary artery (PA) at their origin are plotted for each diagnostic group and compared to the normal range (mean, 5th and 95th centiles). In groups where the aorta was patent and there was unrestricted flow through it, it was normal or close to normal in size. The aorta was dilated in size when there was preferential flow to this vessel, in the setting of pulmonary stenosis

origin of the aorta[16], in cases with concordant ventriculoarterial connections (Groups 1 and 3), but the aortic size was hypoplastic in all. In contrast, the aorta was of good size in all cases in Groups 2 and 4, but the great arteries arose in parallel orientation in these groups. In distinction, the aorta arose to the right of the pulmonary artery in the setting of double outlet, and to the left of the pulmonary artery in those with discordant ventriculoarterial connections (Group 4). In cases with associated pulmonary stenosis, which is common in both double outlet and a discordant connection, the aorta to pulmonary artery ratio will be increased. The degree of relative aortic dilatation will reflect the degree of obstruction to pulmonary flow.

In each case, the mother was referred for detailed fetal echocardiography because of an abnormal four-chamber view noted during an obstetric ultrasound examination. The mean gestational age at detection of congenital heart disease was 26.3 weeks. Only 12 of 24 mothers (50%) were diagnosed prior to 24 weeks' gestation and, therefore, had the option of termination of pregnancy. This is in contrast to a previous series from the UK, in which 68% of cases of all cardiac malformations diagnosed since 1980 were detected prior to 24 weeks, with a mean gestational age in 1992 of 22.7 weeks[2]. This reflects a differing policy between the USA and the UK in offering a complete ultrasound examination, including four-chamber view analysis, as a matter of routine at 18–20 weeks' gestation in the latter country. Although examination of the four-chamber view is a recommended part of AIUM or ACOG guidelines during obstetric evaluation, in general, pregnancies are scanned only for specific indications[19]. As 90% of congenital heart disease occurs in low-risk pregnancies, this policy prevents the identification of major cardiac lesions in the early fetus.

There was a low rate of termination of pregnancy despite the diagnosis of severe disease and in only 3/12 cases diagnosed prior to 24 weeks' gestation did the parents choose this course. This partly reflects the preponderance of socially deprived patients the hospital serves, who are more likely to continue with an abnormal pregnancy. It also reflects the selection of patients for referral to our center, as our results of staged palliation for the hypoplastic left heart syndrome are close to the current best – 65% survival through the third stage. Those patients who are diagnosed with this condition at other obstetric centers in the area and who wish to choose termination are not referred unless they insist on a second opinion.

Chromosomal anomalies were uncommon in the hypoplastic left heart syndrome but occurred in 1/14 (7%). This is similar to the rate of chromosomal anomaly found in a previous group of 161 cases of 4%[2]. In contrast, chromosome anomalies are much more frequent in mitral atresia with double-outlet right ventricle: 1/3 (33%) in this series and 11/60 (18%) in a larger series[2]. In addition, other extracardiac anomalies were found in two further cases of the hypoplastic left heart syndrome which did affect the results of postnatal treatment. Extracardiac anomalies typically associated with isomerism of the right atrial appendages, or the asplenia syndrome, occurred in cases with a common atrioventricular junction (Group 2b). Chromosomal anomalies are extremely rare with this syndrome[20]. No extracardiac lesions were found in either of the cases in Groups 2a or 4. This may be because of the small number of cases in these groups, but chromosome anomalies are particularly rare with atrioventricular discordance (Group 4), and a karyotype is not essential if this firm diagnosis is made.

Forms of congenital heart disease which are associated with an abnormal four-chamber view are preferentially detected prenatally. For example, mitral atresia with ventricular septal defect and dominant ventricle with absence of the left connection (Groups 2a and 4) would be unusual in a series of 100 infants with congenital heart disease[21,22] and yet we have seen two cases of each in our series of 80 fetuses with congenital heart disease. The hypoplastic left heart syndrome, however, is a common lesion, comprising 7–10% of cases of heart disease in infancy[21,22] and 22% in a previous fetal series[2]. The number detected here represents 25% of the total cases diagnosed prenatally in our center.

Of the total group of 24 pregnancies, four sets of parents elected to interrupt the pregnancy; there was one intrauterine death, and ten neonatal deaths. There are nine survivors, with one severely neurologically damaged. There are, therefore, seven currently well babies of 19 continuing pregnancies (37%). These seven face further staged surgery, which has a significant risk. Five patients will have a right ventricular-dependent Fontan circulation and two a left ventricular-dependent Fontan circulation. This form of palliation is likely to have a time-span of 10–25 years[23] before transplantation. These types of heart disease have a profound effect, therefore, not only on the survival but also on the quality of life of affected children. The malformations are clearly detectable in early pregnancy by evaluation of the four-chamber view, which is the easiest cardiac view to obtain and understand. Only half the parents in this series, however, were informed of the findings at a stage of pregnancy when they could have exercised choices in the management of their pregnancy. The ultrasound technology for the detection of malformations is presently available and in place for the general management of pregnancy. Detection of malformations could be increased by improving the standard of fetal ultrasound examination during the second trimester. This can be achieved by simple educational strategies[5].

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