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Prospective Diagnosis of 1,006 Consecutive Cases of Congenital Heart Disease in the Fetus

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Objective. **This report describes our experience with fetal congenital heart disease since 1980.**

Background. **Knowledge and expertise in the diagnosis, management and natural history of fetal congenital heart disease is increasingly demanded by both obstetricians and parents. The analysis of a large series should help the pediatric cardiologist to provide this service.**

Methods. **The notes of 1,006 patients, where a prospective diagnosis of fetal congenital heart disease was made, were reviewed. The reason for referral, the diagnosis made, the accuracy of diagnosis, the fetal karyotype and the outcome of the pregnancy were noted. The cases were grouped into malformation categories, and the spectrum of disease seen was compared with that found in infants.**

Results. **Most fetal cardiac anomalies are now suspected by the ultrasonographer during obstetric scanning. A different incidence of abnormalities is seen compared with that expected in infants. Chromosomal anomalies were more frequent in the fetus than in live births. The accuracy of diagnosis was good. The survival rate after diagnosis was poor because of frequent parental choice to interrupt pregnancy and the complexity of disease.**

Conclusions. **A large experience with fetal congenital heart disease allows the spectrum of disease to be described with accuracy and compared with that in infancy. Knowledge of the natural history of heart malformations when they present in the fetus allows accurate counseling to be offered to the parents. If the trend in parental decisions found in this series continues, a smaller number of infants and children with complex cardiac lesions will present in postnatal life.**

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Toward the end of the 1970s, cross-sectional diagnosis of structural congenital heart malformations became established in children (1,2). Simultaneously, advancing expertise in obstetric ultrasound techniques allowed diagnosis of an increasing array of fetal anatomic defects (3,4). Improvements in real-time imaging around this time prompted several workers to explore the possibility of identifying the structure of the normal fetal heart (5–7). Since then, almost every form of congenital heart disease recognizable by ultrasound in the infant or child has been detected in fetal life (8). In February 1980, with the support of the British Heart Foundation, a referral center for mothers at increased risk for fetal congenital heart disease was established at Guy's Hospital. Between then and February 1993, >1,000 cases of fetal cardiac malformation have been recognized in this center, and these constitute the subject of this report. The aim of the report is to document the changing patterns of referral since the start of the study; to look at the scatter of malformations seen and to compare them with that expected in infancy and to examine the outcome, including parental choice and natural history, in each diagnostic group.

Methods

All mothers seen at the center since 1980 were considered to be at increased risk for fetal congenital heart disease. The high risk cases fell into two groups: those with "maternal" and those with "fetal" high risk factors (Table 1). A total of 10,120 mothers have been seen since February 1980, with the number of referrals increasing each year (Fig. 1). An increasing number of cases of congenital heart malformation have been diagnosed annually (Fig. 2) to a total of 1,006 cases. Diagnosis was made by examination of a videotape recorded at another center in 25 cases (3%). The reasons for referral and the gestational age at referral in the abnormal cases were noted, and their changing pattern was examined. As our experience enlarged, and the high rate of association became evident (9), we increasingly attempted to obtain a fetal karyotype, the results of which were noted where available.

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Table 1. Pregnancies at Increased Risk for Congenital Heart Disease

Maternal factors
Family history of congenital heart disease
Previous child or children affected
Parental congenital heart disease
Maternal diabetes
Exposure to known cardiac teratogens (e.g., lithium, anticonvulsants)
Fetal factors
Extracardiac fetal anomaly
Nonimmune fetal hydrops
Fetal arrhythmia
Abnormal appearance of the heart seen during an obstetric scan

In every case where a heart malformation was found in the fetus, follow-up information was sought. The cardiac specimen was requested in all cases of fetal or neonatal death. Failing this, the report of a local postmortem was obtained wherever possible. Postnatal clinical or investigation findings were obtained in surviving cases.

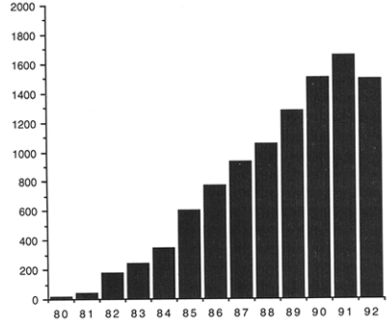
Cases were divided into diagnostic categories that were compared with the pattern of malformations detected in infancy in the New England Infant Cardiac Care Program or the Brompton series (10,11).

Results

Of the 1,006 cases of congenital heart disease diagnosed prenatally, the majority (72%) were referred because of suspicion of a cardiac malformation aroused during an obstetric scan. The reasons for referral in different diagnostic categories are detailed in Table 2. Cases are designated in hierarchic sequence, according to the most "important" lesion present. This is a modification (12) of the proposals of Fyler et al. (10) and Scott et al. (11).

The majority (68%) were diagnosed at <24 weeks of gestation. The average gestational age at diagnosis decreased progressively from 26.5 in 1983 to 22.7 weeks in 1992. Chromosomal analysis was obtained in 412 fetuses, (40% of the total), of which 172 proved positive (42%). In general, those fetuses without a result from chromosomal analysis did not demonstrate associated anomalies suggestive of a karyotypic abnormality; therefore a more correct rate of chromosome anomalies would be closer to 172 of the total, that is, 17%. No further information concerning the diagnosis was available in 120 cases because of parental refusal of a postmortem examination (12%). In all of the remaining cases, follow-up information in the form of a postnatal study or autopsy was available for comparison with the fetal diagnosis, and in 48% of cases, a postmortem examination was performed in our center. Cases are enumerated according to the fetal diagnosis regardless of its accuracy. The accuracy of diagnosis in each category is indicated in Table 2. After counseling, 558 parents chose to interrupt the pregnancy (55%). The outcome after diagnosis in each category is shown in Table 3. In Table 4, the proportion of cases in different diagnostic categories is compared with two infant series (10,11).

Figure 1. Number of high risk referrals per year (1980 to 1992).



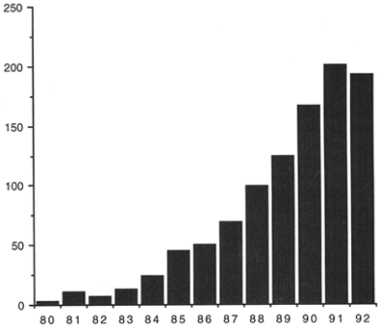


Figure 2. Cases of cardiac anomaly seen per year.

Atrioventricular septal defect. This was the most commonly detected lesion, found in 177 cases. Of these, 19 were thought to have associated right and 48 associated left isomerism of the atrial appendages. Of 112 successful cultures, there were 63 chromosomal anomalies proved in this group, or 35% of the total. Of the added diagnoses, isomerism was found in an additional 11 cases. The diagnoses where an atrioventricular (AV) septal defect was not confirmed were "normal" in two local autopsy reports, inlet perimembranous ventricular septal defects in four and mitral atresia in one.

Hypoplastic left heart. This diagnosis, using the strict criteria of no flow through either the mitral or aortic valves,

Table 2. Reasons for Referral by Diagnostic Category

Diagnosis	No.	Reason for Referral					Follow-Up		
		FH	ECA	FArr	FHyd	?CHD	FUI	AD	INC
Atrioventricular septal defect	177	15	14	7	14	127	154	26	7
Hypoplastic left heart syndrome	161	11	6	1	6	137	127	3	7
Coarctation of the aorta	113	23	18	2	4	66	104	8	32
Tricuspid dysplasia/Ebstein's anomaly	75	6	0	2	1	66	66	6	3
Ventricular septal defect	60	10	17	3	5	25	52	3	5
Mitral atresia	60	5	5	0	0	48	50	0	15
Pulmonary atresia, IVS/critical PS	55	2	0	0	2	49	48	0	4
Tricuspid atresia	45	1	1	0	0	43	43	7	5
Critical aortic stenosis	41	0	1	0	1	39	40	0	0
Cardiomyopathies	41	0	13	1	7	20	37	0	0
Double-outlet right ventricle	33	1	4	1	1	25	26	1	4
Tetralogy of Fallot	31	5	15	1	1	9	27	1	4
Complete transposition	20	6	1	0	0	12	20	0	4
Double-inlet ventricle	18	1	0	0	0	17	17	2	1
Tetralogy with pulmonary atresia	15	2	1	0	0	11	11	1	1
Common arterial trunk	14	7	1	0	0	6	14	0	4
Tumor	13	1	0	0	1	11	13	0	0
Conjoint twins and ectopia cordis	12	0	0	0	0	12	12	5	0
Absent pulmonary valve syndrome	10	0	0	0	0	10	7	0	0
Aortic-left ventricular tunnel	4	0	0	0	0	4	4	3	0
TAPVC	3	2	0	0	0	1	3	2	0
Congenitally corrected transposition	3	0	0	0	0	3	3	0	0
Miscellaneous	(5)						5	0	0
Calcific arterial disease	2	1	0	0	1	0			
Interruption of the aorta	1	1	0	0	0	0			
Mitral stenosis	1	0	0	0	1	0			
Idiopathic right atrial enlargement	1	0	0	0	0	1			

Data are number of cases. AD = main diagnosis confirmed but with additions to central diagnosis; ?CHD = suspicion of congenital heart disease on routine scan; ECA = positive extracardiac anomaly; FArr = positive arrhythmia; FH = positive family history; FHyd = positive fetal hydrops; FUI = follow-up information; INC = incorrect or central diagnosis unconfirmed; IVS = intact ventricular septum; PS = pulmonary stenosis; TAPVC = totally anomalous pulmonary connection. See text for details of each diagnosis.

was made in 161 cases. There were 7 chromosomal anomalies of 40 successful cultures (17%), or 4% of the total. Of the seven cases of differing diagnosis at autopsy, the diagnosis was mitral atresia with double-outlet right ventricle (three cases), mitral atresia with a ventricular septal defect and concordant ventriculoarterial (VA) connections in one and a common AV valve connected to a dominant right ventricle in two. In the remaining case, an apical left ventricular aneurysm was found. In this case, the left ventricle was not markedly hypoplastic, but there was no forward flow through either the aortic or mitral valves. The additional diagnoses made in three cases were a ventricular septal defect in one and left isomerism in two.

Coarctation of the aorta. This diagnosis was suspected in 113 cases from a combination of features, namely, right ventricle larger than the left; the pulmonary trunk dilated relative to the aorta; distal narrowing of the aortic arch and bidirectional or left-to-right shunting at atrial level. There were 33 chromosomal anomalies of 62 successful cultures (53%), or 29% of the total. The defect was confirmed in 72; some other form of congenital heart disease was found to account for the abnormal signs in 14; and a normal heart was discovered in 18.

Tricuspid dysplasia/Ebstein's malformation. There were 75 examples of these conditions. The two diagnoses were grouped together because they overlapped, although there was wide variation in severity in both groups of cases, particularly in the degree of right atrial dilation. There were four chromosomal anomalies of 18 successful cultures (22%), or 5% of the total. The diagnoses where the lesion was not confirmed included an atrial septal defect in one; the heart was said to be "unremarkable" in one local autopsy; and the left atrium and ventricle were said to be markedly dilated in another local autopsy.

Ventricular septal defect. There were 60 cases of ventricular septal defect. There were 29 chromosomal anomalies of 39 successful cultures (74%), or 48% of the total. In four of the five unconfirmed cases, autopsy took place after termination of pregnancy for a chromosomal anomaly. In the fifth case, a ventricular septal defect was clearly defined prenatally, but it had closed at postnatal examination. Additional diagnoses found at autopsy were mitral atresia (1), double-outlet right ventricle (1) and congenitally corrected transposition (1).

Mitral atresia. This diagnosis, which excludes examples of the hypoplastic left heart syndrome, was made in 60

Table 3. Outcome

Diagnosis	CA (%)	TOP (%)	IUD (%)	NND (%)	ChD (%)	S (%)
Atrioventricular septal defect	35	62	20	11	3	37
Hypoplastic left heart syndrome	4	72	5	20	0.6	4
Coarctation of the aorta	29	46	11	10	4	51
Tricuspid dysplasia/Ebstein's anomaly	5	44	14	17	2	38
Ventricular septal defect	48	41	16	15	3	40
Mitral atresia	18	68	6	18	3	11
Pulmonary atresia, IVS/critical PS	5	50	9	7	1	63
Tricuspid atresia	2	53	4	4	6	61
Critical aortic stenosis	0/2	56	2	31	2	11
Cardiomyopathies	2	35	20	15	2	40
Double-outlet right ventricle	12	75	6	6	0	50
Tetralogy of Fallot	27	35	13	16	13	35
Complete transposition	0	40	0	5	0	92
Double-inlet ventricle	0	72	0	1	0	80
Tetralogy with pulmonary atresia	13	40	6	33	13	11
Common arterial trunk	14	50	0	40	30	0
Tumor	0	38	30	0	0	44
Conjoint twins and ectopia cordis	0	58	0	33	0	20
Absent pulmonary valve syndrome	20	30	30	30	10	0
Aortic-left ventricular tunnel	0	75	0	0	25	0
Totally anomalous pulmonary venous connection	30	0	0	33	33	33
Congenitally corrected transposition	0	66	0	33	0	0
Miscellaneous	0	0	40	20	20	20

CA = chromosomal anomaly; ChD = % of total death in infancy or childhood; IUD = % of total spontaneous intrauterine death; NND = % of total resulting in neonatal death; S = % survivors of continuing pregnancies (those pregnancies that were not electively terminated); TOP = % of total choosing termination of pregnancy; other abbreviations as in Table 2.

cases. The aorta arose from the left ventricle in two cases and from the right ventricle in the remainder. There were 11 chromosomal anomalies of 28 successful cultures (39%), or 18% of the total. Of the unconfirmed cases, the lesion was found to be mitral stenosis in two cases, double-inlet ventricle in three, AV septal defect committed to the right ventricle in four, tricuspid atresia in three, complete transposition with ventricular septal defect in one and congenitally corrected transposition in one. The reason for referral was maternal diabetes in two cases.

Critical pulmonary stenosis/pulmonary atresia, intact ventricular septum. There were 55 cases in this group. They were grouped together because the former frequently evolved into the latter as pregnancy advanced. There were 3 chromosomal anomalies of 15 successful cultures (20%), or

Table 4. Comparison of Diseases Seen in Infant and Fetal Series

Diagnosis	Brompton (%)	NERICP (%)	Fetal (%)
Ventricular septal defect	15.4	15.7	5
Complete transposition	10.4	9.9	2
Tetralogy of Fallot	9.9	8.9	3
Coarctation	10.5	7.5	11
HLHS	3.7	7.4	16
Mitral atresia	0	0	5
Critical aortic stenosis	1.1	1.9	4
AVSD	3.9	5.0	17.5
Pulmonary stenosis/atresia	4.9	6.4	5
Atrial septal defect	0.5	2.9	0
TAPVC	3.6	2.6	<1
Cardiomyopathy	2.7	2.6	4
Tricuspid atresia	4.7	2.6	4
Double-inlet ventricle	4.3	2.4	2
Double-outlet right ventricle	3.0	1.5	3
Trunk	2.1	1.4	1.5
Ebstein's malformation	0	0	7
Tumor	0	0	1
Corrected transposition	0.8	0.9	<1
Miscellaneous	11.3	10.4	<1

AVSD = atrioventricular septal defect; Brompton = Royal Brompton Hospital, London; HLHS = hypoplastic left heart syndrome; NERICP = New England Infant Cardiac Care Program; TAPVC = totally anomalous pulmonary venous connection.

5% of the total. The reason for referral was maternal diabetes in two cases. Of the four cases unconfirmed, the lesion was shown to be tricuspid atresia in one, complete transposition with aortic hypoplasia in one and AV septal defect committed to the right ventricle with pulmonary stenosis in one; and in one of twins, thought to have pulmonary stenosis because of a tricuspid regurgitation velocity of 3 m/s, the postnatal echocardiogram was normal.

Tricuspid atresia. This diagnosis was made in 45 cases. Discordant VA connections were also found in 12. There were 2 chromosomal anomalies of 14 successful cultures (14%), or 2% of the total. Of the unconfirmed cases, the lesion was shown to be AV septal defect draining to the left ventricle in five. In three cases, the great arteries, thought echocardiographically to be concordant, proved to be discordantly connected. Coarctation (n = 1), interruption (n = 2) and right isomerism (n = 1) were found in addition in four cases.

Critical aortic stenosis. There were 41 cases in this diagnostic group. One chromosomal anomaly was detected in four fetuses tested, but this was an abnormal y chromosome of questionable significance (0.25%), or 0.2% of the total.

Cardiomyopathies. This was a miscellaneous group of 41 cases of varying types and appearances. There was one chromosomal anomaly of 8 successful cultures (12.5%), or 2% of the total. The diagnosis was confirmed in all cases with follow-up, although in six infants the appearances normalized in the first year of life. Proved underlying diagnoses included renal anomalies in 12, Noonan's syndrome in 2,

toxoplasmosis in 1, parvovirus in 1, maternal diabetes in 1 and sialic acid storage disease in 1.

Double-outlet right ventricle. This diagnosis was made in 33 cases. There were 4 chromosomal anomalies of 18 successful cultures (22%), or 12% of the total. The reason for referral was maternal exposure to diphenylhydantoin in one case. Of four cases unconfirmed, the lesion proved to be corrected transposition in three and common arterial trunk in one. The additional diagnosis of mitral atresia was made in one case.

Tetralogy of Fallot with pulmonary stenosis. This diagnosis was made in 31 cases. There were 9 chromosomal anomalies of 25 successful cultures (36%), or 27% of the total. Of the two cases unconfirmed, the heart was not mentioned in a local postmortem report in one, and the lesion had progressed to tetralogy with pulmonary atresia in the other, although there had been forward flow clearly documented prenatally.

Complete transposition. The diagnosis of concordant AV and discordant VA connections was made in 20 cases. There were no chromosomal anomalies in this group of cases. The reason for referral was exposure to maternal infection in one case. Of four unconfirmed cases, congenitally corrected transposition was proved in three and double-outlet right ventricle in one.

Double-inlet ventricle. This diagnosis was made in 18 cases. There was left and right isomerism in one case each in addition. There were no chromosomal anomalies in this group of cases of three tested. In the one case where the defect was unconfirmed, two very large ventricular septal defects were found. In one case, left isomerism was found in addition, and in another case, thought to have coarctation, an interrupted aortic arch was proved.

Tetralogy of Fallot with pulmonary atresia. This diagnosis was made in 15 cases. There were two chromosomal anomalies of 11 tested (18%), or 13% of the total group. The reason for referral was maternal diabetes in one case. In the one case where the defect was unconfirmed, a common arterial trunk was proved. In one case, AV septal defect and right atrial isomerism were found in addition.

Common arterial trunk. This diagnosis was made in 14 cases. There were two chromosomal anomalies in this group of seven tested (29%), or 14% of the total. Of four cases unconfirmed, three proved to be tetralogy of Fallot with pulmonary atresia and one concordant great arteries, with the aorta anterior and to the left of the pulmonary artery, and a ventricular septal defect.

Tumor. Cardiac tumor was detected in 13 fetuses. There were no chromosomal anomalies associated with this group. One tumor was multicystic and lay in the pericardial cavity. The others were all intracardiac, and nine were multiple. Of the nine cases examined histologically, the cystic tumor was shown to be a teratoma, whereas the others were rhabdomyomas. Three of the four surviving children have signs of associated tuberose sclerosis, although their tumors have regressed in size postnatally.

Absent pulmonary valve syndrome. This condition was detected in 10 cases. There were two chromosomal anomalies of six tested, or 2% of the total group.

Thoracopagus conjoint twins and ectopia cordis. There were seven examples of the former and five of the latter diagnosis. There were no chromosomal anomalies in this group of cases. The primary defect was confirmed in all cases, although the specifics of the intracardiac anatomy were delineated more completely in five of the cases of conjoint twins at autopsy. One child with ectopia is alive. This infant had a small lower sternal defect with protrusion of the left ventricular apex and tetralogy of Fallot, both of which have been surgically repaired.

Aortic left ventricular tunnel. There were four fetuses with this condition. In four cases, aortic regurgitation and left ventricular volume overload were detected, although the correct mechanism for this finding was diagnosed prospectively in only one. Postmortem study at our center took place in all cases and demonstrated the anatomic site of the aortic incompetence as paravalvular because of an aortic left ventricular tunnel. The presence of a ventricular septal defect, suspected in addition in one case, was not confirmed.

Congenitally corrected transposition of the great arteries. The prospective diagnosis of discordant AV and discordant VA connections was made in three cases. There were moderately severe degrees of Ebstein's malformation of the posterior, morphologically tricuspid, valve in all cases. There were no chromosomal anomalies associated with this diagnosis.

Totally anomalous pulmonary venous connection. This prospective diagnosis was made in one case in which a chromosomal anomaly was found in addition. The diagnosis was confirmed by postnatal echocardiography. Two additional cases with right ventricular volume overload were thought to be unspecified examples of heart disease, perhaps coarctation of the aorta, prenatally. The correct diagnosis was made postnatally in both.

Miscellaneous. There are five cases in this group. There were no chromosomal anomalies. There were two cases of calcific arterial disease, presenting with fetal hydrops in the same mother in two consecutive pregnancies. Both were proved by autopsy after spontaneous intrauterine death. There was one case of congenital mitral stenosis in a twin pregnancy. This fetus was hydropic and died soon after birth, when the diagnosis was confirmed. There was one example of aortic interruption, confirmed postnatally at surgery. This infant died. One fetus was seen because of cardiomegaly. There was a normal tricuspid valve with no regurgitation but moderate right atrial enlargement. These findings have persisted postnatally but the child remains clinically well.

Discussion

Since 1980, at the start of our program, fetal echocardiography has changed from a marginal special interest to an

essential part of a pediatric cardiology practice. Before the start of our teaching drive in 1986, the fetal heart was rarely detected to be abnormal at routine scanning. The other reasons for referral listed in Table 1 produced a low yield of positive cases because only 10% of congenital heart malformations will occur in pregnancies selected for these reasons. The concept of four-chamber view analysis in screening for congenital heart disease during routine obstetric scanning immediately widened the potential impact of the technique (13). Thus, since this time, in one study up to 70% of four-chamber view anomalies were detected in a normal screened pregnant population (14). In addition, a decrease in the postnatal detection of conditions, such as the hypoplastic left heart syndrome, can be demonstrated as a result of parental choice for termination of pregnancy after early diagnosis (15). As the expertise of the ultrasonographer extends and improves, and the demands of parents for early prenatal detection of fetal malformations increase, it is likely that in the future, the majority of cardiac connection anomalies will be detected in fetal life. If parent choice continues in the same trend as before, this will lead to a decrease in the number of children with complex cardiac malformations.

Reasons for referral. The most common reason for referral for fetal echocardiography is a family history of congenital heart disease, although only 2% proved to have recurrent disease. Maternal diabetes was associated with mitral atresia (two cases), pulmonary atresia (two cases) and tetralogy with pulmonary atresia (one case). Exposure to anticonvulsants and maternal infection were associated with examples of double-outlet right ventricle and transposition with a ventricular septal defect. In 1986, we initiated an educational program for ultrasonographers and obstetricians, concentrating mainly on encouraging examination of the four-chamber view of the heart during routine scanning. Since this time, the majority of fetuses found to have a cardiac anomaly are referred with a suspicion aroused during the evaluation of this one cardiac section (16). Examination of Table 2 shows that there are some cardiac defects, usually those which are not seen on a four-chamber view, that were detected prenatally because of a family history of congenital heart disease. Among these are coarctation of the aorta, common arterial trunk and complete transposition.

Chromosomal anomalies. Chromosomal anomalies in our series were detected more frequently than they are found in infants: 17% compared with 12.8% found in the Baltimore-Washington Infant Study (17). They were not found in patients with transposition, critical aortic stenosis or in double-inlet ventricles but did occur in almost all other categories. This high rate is mainly related to the selection of patients for referral for fetal echocardiography but also to the increased rate of spontaneous fetal loss in chromosomally abnormal fetuses that would not be detected in postnatal series. The rate of chromosomal defects was expectedly high with respect to AV defects but unexpectedly high with respect to ventricular septal defects, coarctation and tetralogy of Fallot. This emphasizes the importance of chromosomal analysis, not only in all ongoing pregnancies, but also to complete the diagnosis in interrupted pregnancies and to allow accurate counseling about future pregnancies.

Accuracy of diagnosis. The accuracy of diagnosis of fetal cardiac malformations from ~18 weeks of gestation is good and has improved with increasing experience. Table 2 indicates the number of cases in each diagnostic group where the diagnosis was added to or unconfirmed, and the correct diagnosis is detailed in the text. This shows that most incorrect diagnoses were differences that would not significantly affect the prognosis for the child or were diagnoses unconfirmed by a local autopsy. Postmortem reports from other centers were often very elementary in their analysis of the heart. There were several instances where a local autopsy reported the heart to be normal, but when the specimen was obtained by us, this did not prove to be correct. In a few cases, nonetheless, the report of the local autopsy has had to be accepted because the specimen was subsequently destroyed and could not be examined by us. In our overall experience, there were very few false positive diagnoses of congenital heart malformation where none existed. The exception to this is in the diagnosis of aortic coarctation, where we have emphasized previously that a false positive diagnosis can be made in the last weeks of pregnancy (18). The accuracy of diagnosis was not significantly different when made by review of a videotape recorded at a remote center.

Comparison with lesions seen in infancy. A clear difference in the proportion of individual anomalies between our fetal and published infant series can be seen in Table 4. In the fetus, the detection rate is much higher of the hypoplastic left heart syndrome, Ebstein's malformation, AV septal defects, mitral atresia and, to a lesser extent, critical aortic stenosis. This is because these are all lesions readily detectable during the screening four-chamber scan. In contrast, the rate of detection of ventricular septal defects, complete transposition and tetralogy of Fallot is lower than in infancy because these lesions are not readily detectable on a routine scan.

Outcome of pregnancy. Parents are counseled concerning the type of cardiac anomaly present and their options explained in a nondirective manner. The options will depend on the gestational age at diagnosis and the presence or absence of other fetal anomalies. The prognosis and surgical options available were modified as the natural history in each diagnostic group became more clear with time and as surgical results changed in individual categories. Parents were supported in their decision whatever their choice. The rate of termination of pregnancy can be seen in the different diagnostic categories in Table 3. It is high in lesions such as the hypoplastic left heart syndrome, double-inlet ventricle and mitral atresia. Despite a good prognosis offered for complete transposition in recent years, some parents will still choose to interrupt the pregnancy. The "natural" history of examples of congenital heart disease seen in the fetus is illustrated to some extent by the proportion of survivors of

continuing pregnancies in Table 3 (last column [5]), but in some examples, the least favorable cases were removed by termination of the pregnancy. In addition, in many cases surgery has affected the outcome. The results in reality indicate only the short-term prognosis because over half of the fetal cases have been followed up <3 years. Thus, the survival rate is good for cases of tricuspid atresia, pulmonary atresia with intact septum and double-inlet ventricle because of the success of palliative surgery, although this rate may be expected to decrease with longer follow-up (19). The survival rate is very poor in the hypoplastic left heart syndrome, common arterial trunk, critical aortic stenosis, mitral atresia and pulmonary atresia with a ventricular septal defect. Overall at the present time, about half the parents chose to interrupt the pregnancy where the diagnosis of a major cardiac malformation is made. This number may increase if the gestational age at diagnosis continues to decrease. On a national basis in the United Kingdom, however, <20% of parents are offered this choice because a diagnosis of congenital heart malformation is not identified prenatally. This choice is even less available in the United States, as routine scanning is not generally practiced, and even where it is, the detection of all major malformations is low (20). Were the four-chamber screening program effectively extended to all pregnant women at 18 weeks, and the pattern of parental choice seen in this series continued, this would have profound effects on the future practice of pediatric cardiology.

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