

Personal practice

Diagnosis of fetal cardiac abnormalities

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The feasibility of cross sectional imaging of the structure of the fetal heart was first described in 1980.^{1–3} Since then fetal echocardiography has been carried out in many pregnancies that are at increased risk of congenital heart disease, and more recently the study of one section of the fetal heart has been included in routine obstetric ultrasound examinations.⁴ These studies have yielded surprising results about the incidence, range, and natural history of congenital heart disease that may be seen prenatally. Also the pattern of recurrence of congenital heart disease within family groups does not seem to conform to previously accepted genetic theories.

The mothers who are at increased risk of fetal heart disease include: those with a family history of congenital heart disease; those with diabetes; or those in whom fetal hydrops, arrhythmia, or an extracardiac anomaly has been found. Our most important source of detection of cardiac anomalies in recent years comes from apparently normal pregnancies in which the obstetrician has noticed an abnormal appearance of the heart on a routine scan.

The idea of prenatal cardiac screening came from a French study first reported in 1985 (Fermont L, De Geeter B, Aubry MC, Kachaner J, Sidi D. A close collaboration between obstetricians and cardiologists allows antenatal detection of severe cardiac malformations by 2D echo. Abstract, Second World Congress of Pediatric Cardiology, New York, 1985). A small group of obstetricians was taught to visualise and understand the normal appearance of the four chamber view of the heart and incorporate this into their routine scan (fig 1). When difficulty was experienced in seeing the normal four chamber view the case was referred to a paediatric cardiologist for further evaluation. During a two year period, 20 000 pregnancies were screened, and 39 serious anomalies were detected. This is in accord with the expected figure of 2/1000 that might be detected by this view alone from analysis of data from infants.⁵ Screening for congenital heart disease is therefore possible, and it is now being increasingly practised in

the United Kingdom. Our success has been less dramatic than the French, because most routine scans in this country are done at 16 weeks' gestation, whereas in France they are done at 20–24 weeks when the fetal heart can be seen more easily. Also some obstetric units in the United Kingdom are handicapped by poor ultrasound equipment or limited time available for adding extra work to the schedule. These problems are slowly being resolved, and we currently have a research programme that is trying to improve the quality and reliability of screening. Although there are still a substantial number of serious anomalies missed by this programme the referral pattern over the years has changed enormously as a result of teaching. Fig 2 shows the increasing number of anomalies detected since 1980, and the proportion of these diagnosed from routine scans.

Although the overall frequency of 2 anomalies/1000 detected by the screening programme is consistent with the expected rate from postnatal

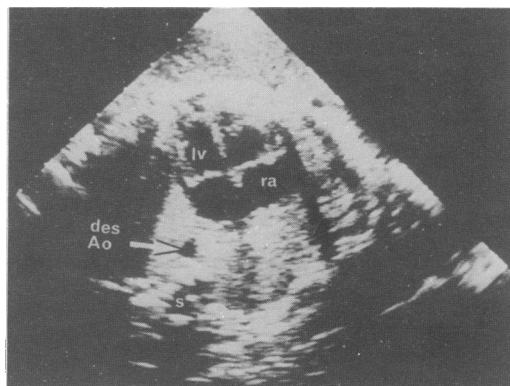


Fig 1 *Normal four chamber view of the fetal heart showing two equally sized atria and ventricles. The heart should occupy about a third of the thorax and the atrial and ventricular septa should meet the atrioventricular valves in a cross at the centre of the heart.*

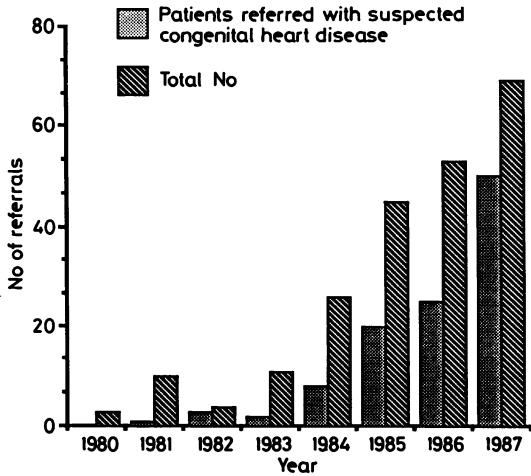


Fig 2 Total number of detected abnormalities continues to rise, but an increasing proportion is suspected before referral.

studies, the range of congenital heart disease is different.^{6,7} Fig 3 shows a comparison of the diagnostic categories of cases seen prenatally and those reported postnatally. Severe forms of heart disease such as hypoplastic left heart syndrome predominate in prenatal life, and forms of heart disease that are unusual postnatally commonly present in fetal life. Tricuspid dysplasia and the related Ebstein's malformation (fig 4) make up 10% of the anomalies seen in our series. In this condition there is gross tricuspid regurgitation and right atrial dilatation, and there may be associated anatomical or functional pulmonary atresia. The prenatal cardiomegaly restricts lung development resulting in severe pulmonary hypoplasia. Such a neonate is impossible to resuscitate, and dies soon after delivery. Early death accounts for the fact that this condition is rarely encountered in paediatric cardiological practice.

The natural history of some forms of fetal heart disease is unexpected. Many conditions, including hypoplastic left heart syndrome, atrioventricular septal defects, and critical aortic stenosis may present with intrauterine heart failure (fetal hydrops). Spontaneous intrauterine death occurred in 14% of our total series of cases detected prenatally, and this does not include those cases where termination of pregnancy took place in anticipation of intrauterine death. There is a particularly high rate of spontaneous fetal loss in cases of heart disease associated with chromosomal anomalies; the overall incidence in our series of cardiac malformations is 20%. The finding of unusual forms of prenatal heart

disease and the high rate of intrauterine death suggest that the incidence of congenital heart disease in early fetal life may be significantly higher than that in postnatal life.

Some abnormalities change or progress during

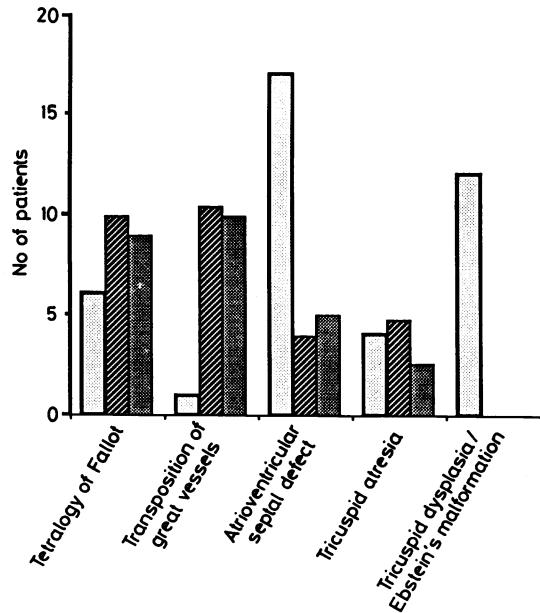
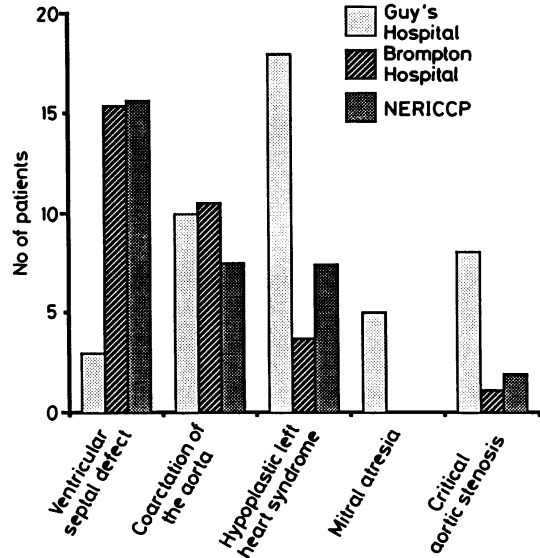


Fig 3 The range of disease seen in prenatal life in the Guy's unit compared with the scatter seen in infancy reported from the Brompton Hospital, and the New England Regional Infant Cardiac Care Program.

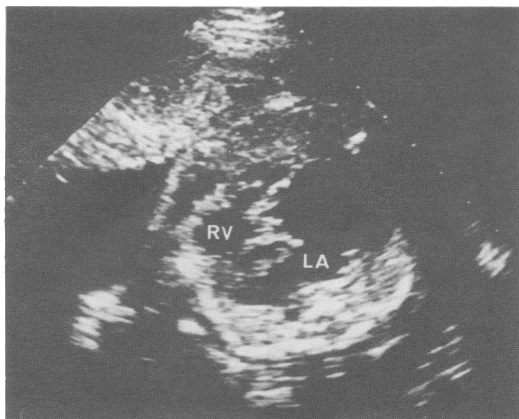


Fig 4 The heart is enlarged relative to the thorax because tricuspid valve dysplasia and regurgitation causes atrial dilatation, mainly on the right.

prenatal life. For example, a patient with coarctation of the aorta may develop severe hypoplasia of the aortic arch; the pulmonary arteries in a patient with tetralogy of Fallot may become more hypoplastic; and tumours may enlarge as pregnancy progresses. In several cases the pulmonary valve and right ventricle have looked normal in the early fetal study but serious pulmonary stenosis has developed by full term.

The recurrence of congenital heart disease within a family group seems to depend on the lesion found in the index case, but recurrences do not occur in patterns consistent with generally accepted genetic theories.⁸ The overall rate of recurrence is close to the predicted 1/50 but some conditions recur more often than others. The hypoplastic left heart syndrome, coarctation of the aorta, common arterial trunk, and conditions associated with atrial isomerism, are examples of malformations that may recur at a rate of as high as 1/10 when a previous child in the family has been affected. The recurrence rate when a parent is affected is still not clear, but is of the order of 1/10.

The advantage of detection of fetal congenital heart disease

There are several advantages to the prenatal detection of fetal congenital heart disease. The first is to give the parents the option of termination of pregnancy when severe heart disease with a poor long term outlook is found early in gestation. Despite advances in paediatric cardiac surgery there is still a high mortality for many conditions in childhood, before, during, and after operative treat-

ment. The information provided about the heart should be as accurate and objective as possible. The echocardiographer must be aware of the strong association of some forms of heart disease with extracardiac and chromosomal anomalies and take steps to ensure that the parents make decisions with all the possible information available to them. Fig 5 shows an atrioventricular septal defect, which led to the diagnosis of Down's syndrome in the fetus of a young mother. Most parents when faced with the facts of the individual condition found in their fetus choose termination. In our series 75% have taken this step when the diagnosis has been made before 28 weeks' gestation. If screening becomes more widespread there are enormous potential implications for the future practice of paediatric cardiology. Most parents have thought of the possibility of fetal abnormality and how they might react if it happened to them. They already have established views on the morality of abortion in the context of their own lives. Those who have previous experience of congenital heart disease in the family, especially if that child died, are usually particularly anxious to avoid repeating the experience. My attitude is that this decision belongs to the parents alone and that they should be supported whatever decision they make.

Theoretically, when a prenatal diagnosis of heart disease is made and antenatal care is transferred to a unit with combined facilities for obstetrics and paediatric cardiology, the infant should have the optimum chance of survival. Our results, however, do not bear this out, mainly because of the complexity and severity of abnormality selected for presentation for fetal echocardiography, and the



Fig 5 A large defect in the atrial and ventricular septa at the crux of the heart. There is a common atrioventricular valve, which is open in this diastolic frame.

incidence of multiple associated extracardiac anomalies. In our series, of 109 babies born alive only 46 survived the first month of life. Of the total series, there are only 22 normal children still alive with either corrected or readily correctable lesions.

Psychological assessment of mothers seen in our department suggests that a mother is emotionally better able to cope with the infant when she has had time to prepare herself prenatally for the problems to be encountered in neonatal life than the mother who learns of her newborn baby's heart disease in the postnatal ward. Many doctors feel that if a mother finds out about a fetal abnormality during pregnancy it will interfere with the experience of pregnancy, but they underestimate most mothers' subconscious and ever present fears of fetal anomaly. Most mothers would elect to have every possible test for the detection of fetal abnormalities if there was no risk attached to the test. The demand for fetal echocardiography has risen from 35 high risk pregnancies in 1980 to over 1000 patients in 1987. The few mothers who do not wish prenatal diagnosis of any kind are able to opt out if they wish.

A further advantage of fetal echocardiography is our increased knowledge of the prevalence and natural history of congenital heart disease in fetal life. This raises the possibility of intervention during prenatal life. It is possible to insert a needle safely into the umbilical cord and even directly into the heart.^{9 10} Will it ever be possible to dilate an arterial valve or burn it with a laser at the end of a catheter prenatally? Would this change the outcome in conditions with a poor prognosis, such as critical aortic stenosis or pulmonary atresia with an intact ventricular septum? Some forms of hypoplastic left heart syndrome have an intact foramen ovale. Would opening this change the course of left ventricular development? These are futuristic concepts but perhaps not so impractical as they first seem. In the United States the time between prenatal diagnosis of the hypoplastic left heart syndrome and delivery is already being used to advantage by seeking a suitable donor heart for transplantation. If this proves to be successful in the long term it may be an avenue to explore in this country. Although I feel that it is fraught with medical, social, and financial problems, some parents are willing to submit their child to any procedure that may give a possible chance of survival. The maximum possible freedom of choice, based on clear, accurate, and unbiased information should be available.

Cardiac function in prenatal life

Most heart disease in infancy and childhood is

structural rather than functional. Similarly, disturbed anatomy of the heart is the most common disorder to look for prenatally. Functional anomalies, however, do occur. As in postnatal life, cardiac function can be assessed by looking for peripheral signs of cardiac failure or, more directly, by echocardiography. Cardiac failure in utero is manifest not only by ascites and cardiomegaly as measured by the cardiothoracic ratio, but also by pericardial and pleural effusions and skin oedema. These signs can also be caused by non-cardiac lesions.

Cardiac function can also be assessed by Doppler examination of the intracardiac valves. The passive filling phase of the atrioventricular valve tracing will give some indication of ventricular compliance. Atrioventricular valve regurgitation is commonly associated with fetal hydrops. Using a product of the mean velocity of valvar flow and the area of the valve orifice, the volume of blood flow expelled by each side of the heart can be calculated.¹¹⁻¹³ Although this estimation is sometimes inaccurate, it is reasonably reproducible. The output from the right heart is always slightly greater than the left, with a ratio of about 1.3:1. Cardiac output is well maintained until late in the course of fetal compromise¹⁴ but a fall in output indicates a poor prognosis.

Treatment of the fetus

Some possible future invasive interventional techniques have already been mentioned. At present, treatment is confined to giving drugs to the mother to improve the condition of the fetus, but this is mainly for fetal tachycardias.¹⁵ The type of arrhythmia must be documented by M mode echocardiography before treatment is started. An atrial tachycardia without evidence of cardiac failure will usually be converted to sinus rhythm with digoxin alone. For cardiac failure, digoxin and verapamil or flecainide have been successfully used. Premature delivery of a hydropic fetus should be avoided as these infants have a particularly high incidence of necrotising enterocolitis. Once the tachycardia is under control the hydrops can resolve and delivery can take place nearer full term.

In summary, ultrasound examination of the fetal heart can accurately diagnose structural and functional heart disease. It has provided extensive information on the natural history of many forms of congenital heart disease and on normal haemodynamics. There are many possibilities for future developments and applications of the technique.

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