

Annotation

Changing impact of fetal diagnosis of congenital heart disease

Introduction

Cardiac abnormalities are an important cause of death in infancy and childhood. In a significant number of cases the cardiac lesion is not recognised during life.¹ Detailed descriptions of the cross sectional appearance of the normal human fetal heart have been published by several authors in 1980.²⁻⁴ The heart can now be studied in the early mid-trimester fetus between 14–18 weeks of gestation and most major malformations of the heart can be diagnosed during fetal life.⁵ It is still the severe end of the spectrum of congenital heart disease that is detected prenatally,⁶⁻⁸ so the outlook for many cases remains poor and parents often have the option to interrupt the pregnancy following prenatal diagnosis. Concern has been voiced about the effects that prenatal diagnosis will have on paediatric cardiology services. The impact of fetal cardiology is clearly an issue that needs to be addressed.

What can be detected prenatally?

Not all forms of fetal cardiac malformations can be detected. It is particularly important that the parents of the baby being scanned understand this and are made aware of the limitations of the procedure. The main cardiac lesions that can be detected during fetal life are outlined in table 1. The lesions are divided into those that would usually be associated with an abnormal four chamber view of the fetal heart and those associated with a normal four chamber view but which have abnormalities of the great arteries. The abnormalities associated with an abnormal four chamber view are potentially detectable by four chamber view screening during routine obstetric ultrasound examination, and extension of this type of screening will lead to a significantly increased number of babies with major cardiac defects being diagnosed prenatally.

What cannot be detected prenatally?

Table 2 lists the lesions that cannot be detected in fetal life. The fact that the fetal circulation is different from the postnatal circulation accounts for the inability to detect some of these, such as a persistent arterial duct and a secundum atrial septal defect; the arterial duct and foramen ovale are naturally patent at the time of performing fetal cardiac scans. The resolution of the ultrasound imaging systems makes it difficult to detect some types of ventricular septal defects, but some of these could easily be overlooked even on postnatal echocardiography were it not for the clinical signs of a ventricular septal defect. The milder forms of obstructive lesions of the aorta and pulmonary artery can develop later and there may be no signs of obstruction during fetal life. In experienced centres many of the lesions that are overlooked during fetal life are considered to be more minor forms of congenital heart disease and these are usually treatable. However, this aspect cannot be dismissed lightly as some of the lesions may require either surgical or catheter intervention, a major event for the parents, particularly if they have been reassured during pregnancy that the heart is normal. It is

vital, therefore, to outline to the parents the types of lesion that can be overlooked and to indicate that treatment may be required in some instances.

What are the effects on paediatric cardiology practice?

The concept of prenatal screening for congenital heart disease was introduced in the UK in 1986⁹ following the results of a French study (Fermont L, *et al.* Abstract presented at the Second World Congress of Paediatric Cardiology, 1986). Subsequently, a concentrated screening programme was set up in 10 obstetric centres in the South East Thames Region in 1988.¹⁰ This study showed that in over 2½ years, 77% of all the cardiac abnormalities associated with an abnormal appearance of the four chamber view of the heart, were detected prenatally in the 10 obstetric scanning centres, following appropriate training of the staff involved in scanning. In 67% of cases, the heart defects were detected as a result of four chamber screening and in a further 10% associated high risk factors prompted referral for detailed fetal echocardiography. In this particular study 67% of the pregnancies, in which a major cardiac defect had been diagnosed in the baby, resulted in a termination.

The increased rate of termination has raised concerns about the future practice of paediatric cardiology, should these results be extended to all obstetric units nationwide. However, a study from the Northern Region, which examined the prevalence and spectrum of neonatal and infant congenital heart disease in a defined population, has shown that a fetal cardiology service will not have any significant impact on paediatric cardiology practice in that region.¹¹ This may also prove to be the case at a national level as the success of prenatal screening for congenital heart disease is related to the expertise and experience of the ultrasonographers performing the scans,⁶⁻¹²⁻¹⁵ and this is not uniform

Table 1 Lesions associated with four chamber view

Lesions associated with abnormal four chamber view	
(a) At the venous-atrial junction	Total anomalous pulmonary venous drainage*
(b) At the atrioventricular junction	Mitral atresia Tricuspid atresia Atrioventricular septal defect Ebstein's anomaly/tricuspid valve dysplasia
(c) At the ventriculo-arterial junction	Aortic atresia Pulmonary atresia with intact interventricular septum Critical aortic stenosis Critical pulmonary stenosis Coarctation of the aorta*
(d) Other	Ventricular septal defects Cardiomyopathy
Lesions associated with a normal four chamber view	
	Transposition of the great arteries Double outlet right ventricle Tetralogy of Fallot Pulmonary atresia with a ventricular septal defect Common arterial trunk Absent pulmonary valve syndrome

Table 2 Lesions overlooked prenatally

Persistent arterial duct
Secundum atrial septal defect
Milder forms obstructive lesion of great arteries (milder forms aortic stenosis, pulmonary stenosis and coarctation of the aorta)
Some forms ventricular septal defects

Table 3 Termination rate for cardiac abnormalities

Year	Total number of fetuses with CHD	Number resulting in termination (%)
1990	166	100 (60)
1992	203	123 (61)
1993	185	116 (63)
1994	173	93 (54)
1995	183	64 (35)

across the country. Currently, obstetric units vary considerably in their ability to pick up cardiac lesions. Some obstetric units are now detecting most abnormalities associated with an abnormal four chamber view and also some great artery abnormalities; other units have a much poorer rate of detection of even the four chamber abnormalities.

In the past five years nearly all the paediatric cardiology centres in the UK have recognised the importance of a fetal cardiology service and have either developed such a service, or are in the process of doing so. As this service has become available nationally, we may be able to assess the real influence of prenatal screening for congenital heart disease on the overall workload of paediatric cardiology centres in the near future. The British Paediatric Cardiology Association has set up a multicentre project to look at the impact of fetal echocardiography at a national level. This study is still ongoing and results are not available as yet. The results of this study are likely to show that, overall, prenatal detection has little influence over paediatric cardiology practice, because there is such a wide variation between obstetric scanning units. When, however, the effects of a busy fetal cardiology unit on the workload of the paediatric cardiology unit in the same centre are evaluated, a different view emerges of the impact of such a service.

Impact of a fetal cardiology service

Between 1200 and 1500 pregnant women are scanned each year in the fetal cardiology unit at Guy's Hospital, London. All are referred because they are considered to be at high risk of having a child with congenital heart disease.¹⁶ The number of fetal cardiac abnormalities detected in this group is in the region of 200 each year. Interestingly, both the total numbers scanned and the numbers of abnormalities detected have remained fairly constant despite market changes in the health service and the fact that more centres offer a fetal cardiology service (fig 1). Fetal cardiology service is of necessity consultant-led with support from junior doctors and technicians. With the expansion of fetal cardiac services nationwide, the normal workload of paediatric cardiologists has increased accordingly. This will eventually stretch the available services to their limits, if it has not already done so. Trusts will need to take this into account in their future allocation of funds and provision of services. A structure will have to be formed in a select few units to provide intensive and in-depth training of future fetal cardiologists.

One of the management options following the prenatal diagnosis of a fetal cardiac abnormality is to interrupt the pregnancy, and it has been the uptake of this option that has caused concern to paediatric cardiologists about their future practice. However, the termination rate for cardiac abnormalities is not a constant feature, as shown by table 3. An increasing number of parents are currently electing to

continue the pregnancy. This has significantly increased the workload, not only in the fetal unit but also in the paediatric cardiology/cardiac surgical unit and the neonatal units.

Rather than decrease the workload of the paediatric cardiology unit, the fetal cardiology service has had the opposite effect. The number of babies delivered at Guy's Hospital following prenatal diagnosis of congenital heart disease has gradually increased over the past three years. In 1993 31 babies were delivered at Guy's Hospital following the prenatal detection of a cardiac abnormality. In 1994 there were 59 and in 1995 there were 83. This has also had an effect on the neonatal unit. In 1993 10% of the total admissions to the neonatal unit were after prenatal diagnosis of congenital heart disease, and in 1994 and 1995 this increased to 17% and 20%, respectively. In the same time period (1993–95) there has been a 35% increase in the number of surgical operations.

An increase in workload seen at Guy's Hospital is not likely to be mirrored in other units. In some congenital heart defects, such as total anomalous pulmonary venous drainage and the hypoplastic left heart syndrome (when staged surgery is a realistic option in the latter defect), it is clearly appropriate for the baby to be delivered in the obstetric unit linked to the paediatric cardiology and cardiac surgery units, to ensure optimal treatment. In other less complex defects, such as duct dependent lesions, it can be argued that the baby could be delivered in the local obstetric unit, started on prostaglandin infusion, and then transferred to the paediatric cardiology unit. This has the potential but important disadvantage of separating the baby from its mother. For this reason, when offered the possibility of delivery and cardiac treatment on the same site at Guy's Hospital or the alternative of delivery at the local hospital, most parents choose the former option. Some of the relatively simpler defects do not need transfer of care and can be managed in the local hospital. Clearly, the parents need to be given objective information about these options, and overriding consideration needs to be given to their views. When I have offered parents these choices and explained the disadvantages of long distance travelling to Guy's Hospital, the parents have been least concerned about that aspect and most concerned about being present with the baby.

Changes in practice

There are several reasons for the changes that have occurred in fetal cardiology practice over the past few

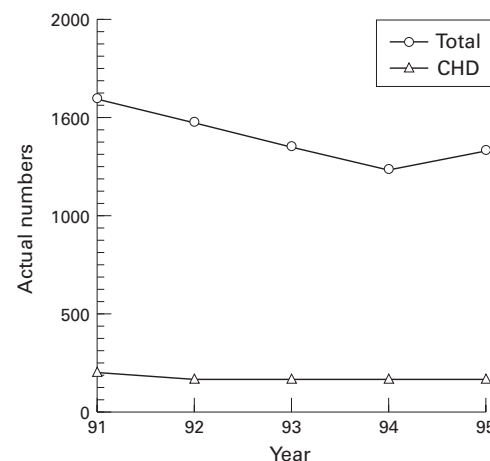


Figure 1 Total numbers of pregnant women scanned each year from 1991–95 and the numbers of structural cardiac abnormalities detected. CHD: numbers of pregnancies with structural heart defects detected in the fetus.

years. Increasing experience of ultrasonographers in obstetric units has improved the detection rate of some of the correctable forms of congenital heart disease such as transposition of the great arteries. Although the detection rate for the great artery abnormalities remains poor, overall, improvement has been steady, if gradual. There have also been advances in surgical techniques, improving the outlook for some treatment options, such as the arterial switch procedure for transposition of the great arteries, or new treatment options for some conditions, such as the Norwood procedure for the management of the hypoplastic left heart syndrome. Over the past few years the extra uterine survival rate has also improved. This probably reflects the improvement in detection rates of correctable lesions and also the changes in surgical practice. The survival rate of the continuing pregnancies seen between 1980–93 in our unit was 41%.^{9 17} Of the pregnancies that continued in 1994 and 1995, the current survival rate is 61%. Although these children are still young and some could still have clinical problems, there does seem to be a notable improvement in the numbers surviving.

Caution is needed when offering certain options to parents during prenatal counselling, but parents can make an informed decision based on the results of treatment currently available. This is a difficult area. The results of the local unit dealing with the patients should be presented, but there is a danger that clinicians will present the best national or worldwide results of a particular treatment to parents, which may imply a better outcome than if the local results had been presented. This can then skew the advice. The provision of adequate information about the problems and results of available treatment is vital in all cases. Most parents require continuing support and are likely to need more than one consultation in order to absorb and understand the implications of the ultrasound findings. Information leaflets and contact with other parents who have had a child with a similar problem are invaluable. We now have the services of a full time nurse counsellor, who is present during consultations with the parents and who forms a crucial part of the service we

offer. The counsellor provides continuing support for the parents throughout the remainder of pregnancy, and in many cases, after delivery.

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