

# Fetal cardiac anomalies

## Diagnosis and management from the perspective of the paediatric cardiologist



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**Congenital heart disease is a leading cause of morbidity and mortality in infancy. Congenital malformations account for about one quarter of all infant deaths and one third of these are of infants with cardiac abnormalities.<sup>1,2</sup>**

Developments in ultrasonic imaging techniques allow the visualisation of the fetal cardiac anatomy and the detection of structural and functional anomalies of the fetal heart as early as 17 to 18 weeks gestation. (Some papers report diagnoses made earlier than this, but in our hands, accurate diagnosis cannot be made reliably in the majority of women until about 18 weeks.)

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The detection of a fetal cardiac anomaly demonstrated early in pregnancy has serious repercussions. Decisions need to be made regarding:

- 1) Whether to continue the pregnancy;
- 2) Whether chromosomal diagnosis is indicated;
- 3) Whether in utero intervention (balloon valve dilatation, maternal drug administration etc) should be considered;
- 4) Whether the baby should be delivered early; and
- 5) Whether the site of delivery of the baby needs to be changed taking into account the likely early neonatal management and facilities available at that site.

Delay in the diagnosis and inter-hospital transfer of a sick neonate are significant factors contributing to the morbidity and mortality of congenital heart diseases.<sup>3,4,5</sup> Parental counselling is very much more productive if diagnosis is made antenatally than when a major cardiac anomaly is identified postnatally: the mother is often exhausted after labour or a caesarean section and may be unable to deal with difficult concepts regarding the baby's cardiac condition. Clearly, some patients will benefit from antenatal consultation with a paediatric cardiologist. In this article, I will try to clarify who might benefit from consultation and what can reasonably be expected from this consultation.

*'Fetal interventions such as balloon valvotomy may well become more commonplace and with improvements in technology, more accurate and earlier diagnoses are likely to be possible.'*

Data presented in this article are drawn from the database of the New South Wales Fetal Cardiac Service. This is staffed by four paediatric cardiologists based at the two Children's Hospitals in Sydney and include Dr Steve Cooper, Dr David Murphy, Dr Megan Sherwood and Dr Gary Sholler. The data were drawn from a random year (2007) and correlated and analysed by our cardiac fellow, Dr Ramesh Parmar. A summary is presented in Table 1.

**Table 1.**  
**Indications for fetal echocardiography**

Indications	Proportion of Total (%)	% abnormality
abnormal fetal heart	41.0	75.3
arrhythmia	5.2	42.3
chromosomal aberration	1.8	44.4
diabetes mellitus	6.5	12.5
maternal drug exposure	1.2	0
extracardiac anomaly	4.4	40.9
family h/o CHD	4.8	16.6
increased nuchal translucency	15.5	19.5
maternal autoimmune disease	0.6	0
maternal CHD	4.0	10.0
paternal CHD	0.4	0
previous fetal death	0.2	0
previous pregnancy with CHD	12.0	9.5
twin to twin transfusion syndrome	1.6	28.6

It is clear that identification of a structural abnormality on screening is likely to correlate with a positive diagnosis. The influence of other risk factors such as family history, chromosomal anomalies and thickened nuchal translucency are listed. I do not have space to discuss each indication for referral and each anatomic diagnosis in this article, but will instead deal with those lesions in which the indication for referral is less clear-cut or where I feel there is a specific benefit for referral.

### Atrial septal defects (ASDs)

All fetal hearts should have a patent foramen ovale (PFO), which in some cases may be quite large. It is seldom possible to be sure that a child has a true secundum ASD in utero. For this reason, I do not think referral is necessary, either because the PFO looks large

or because there is a family history of ASD. The primum ASD is different. It is low in the atrial septum adjacent to the atrioventricular (AV) valves, which are at the same level as each other. The left AV valve is usually cleft and this may be associated with mitral regurgitation. While this lesion will not cause problems perinatally, its recognition is important as it may be associated with Down syndrome and will require repair later.

## Ventricular septal defects (VSDs)

Muscular defects are common and typically small, often evident only on colour Doppler (CD) examination. The defects may be large or multiple and their size influences their postnatal significance. Perimembranous (PM) VSDs are more challenging. The PM region is very thin and on the two dimensional images will often look like a VSD, simply due to 'echo dropout' in the area. It is thus often difficult to be sure about these PM VSDs even with the use of CD. It is sometimes necessary to wait until after delivery of the baby to clarify the situation. The outlet VSD is probably most important in the fetal context. This VSD is under the arterial valve, usually with some override. It raises the suspicion of Tetralogy of Fallot, pulmonary atresia with VSD, truncus arteriosus, transposition of the great arteries (TGA) with subpulmonary VSD (Taussig-Bing anomaly) and various more complex lesions. Identifying an outlet VSD is thus important, both because of the related arterial abnormality and because many of these lesions have chromosomal abnormalities, particularly the Velo-cardio-acial syndrome (VCFS or Catch 22 syndrome).

## Ventricular asymmetry

It is not uncommon to find that the left ventricle (LV) is smaller than the right ventricle (RV) on screening. While variations are sometimes within the normal range, the left side may be disproportionately small and the right side dilated. The commonest pathological explanation for this is a left-sided obstructive lesion, particularly aortic coarctation. This may be associated with aortic or mitral valve abnormalities of varying severity. Imaging the distal arch well enough to diagnose coarctation may be technically difficult and the diagnosis should be considered even if the arch cannot be seen well. I have found that the prognosis for LV function postnatally is generally good if the LV is of similar length to the RV, even if it looks much narrower, but if the LV apex is significantly short of the RV apex, the prognosis is more guarded. This is particularly seen in the context of aortic stenosis rather than coarctation. Diagnosis of arch or aortic obstruction is important as postnatal management usually involves some intervention and the site of delivery chosen needs to be able to deal with this. Also, many children with an interrupted arch and some with coarctation will have VCFS, while some will have Turner syndrome. A less common cause for this pattern of ventricular morphology is premature closure of the PFO. In this situation, the right side is large and often inappropriately hypertensive, with a small and thin-walled LV. Postnatally, the baby will often have low output, but with appropriate support will usually recover.

## Transposition of the great arteries (TGA)

TGA is an extremely important condition to diagnose. It is frequently missed. The main diagnostic clue is the fact that the aorta and main pulmonary artery (MPA) do not cross each other as they leave the heart. The MPA, arising from the LV, will also be seen to bifurcate and the arch (identified by having brachiocephalic vessels arising from it) will come from the RV. The MPA is generally larger than the aorta in the normal heart. In TGA, the vessel arising from the LV is often larger than the vessel arising from the RV and although there

are other explanations for this, TGA should be considered. The reason that it is imperative to identify TGA is that a small number of these children will be extremely ill immediately after birth and may die before a balloon septostomy is able to be performed. The appearance of the PFO just before delivery is not helpful. Antenatally, the PFO has right to left flow which tends to hold it open. After birth the left atrial pressure rises and the foramen closes. Thus, a PFO that looks quite large antenatally may be tiny after birth, allowing a minimal left to right shunt. It does not matter how much blood is passing from aorta to MPA via the PDA, if there is no corresponding left to right atrial shunt, the baby will be in trouble. Having a paediatric cardiologist on site and prepared for an urgent balloon septostomy may avert a potential disaster in a relatively simple manner.

## Intracardiac echogenic foci (IEF)

These lesions are commonly found on the routine antenatal scan. They are usually small (1 to 2 mm) and located on the mitral valve support apparatus, usually at or just above the tips of the papillary muscles. Multiple lesions may be associated with an increased incidence of a chromosomal disorder such as Down syndrome, but the IEFs themselves do not have any pathological significance and do not affect cardiac performance in the short or long-term.<sup>6</sup> We do not recommend paediatric cardiac referral unless the baby has Down syndrome.

## Summary

Fetal cardiology is an exciting field with potential for significant growth in coming years. Fetal interventions such as balloon valvotomy may well become more commonplace and with improvements in technology, more accurate and earlier diagnoses are likely to be possible. The relationship between the screening team and the paediatric cardiologist is important. Obviously, all fetal studies do not require the involvement of a paediatric cardiologist, both because of manpower issues and because it is preferable not to alarm the parents inappropriately. Changing the site of delivery and changing the obstetrician late in the pregnancy is also disruptive and should be avoided if possible. Hopefully, with appropriate communication and experience, babies with significant heart disease can be identified and managed appropriately while the vast majority will continue to be treated normally.

## References

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