

Indications for Fetal Echocardiography

The prevalence of congenital heart disease in live-births varies between 4 and 11 cases per 1000 depending on the methods of ascertainment. There are several categories of pregnancy where there is an increased risk of congenital heart disease, when it is appropriate to refer the patient for a targeted fetal echocardiogram. Risk factors can be divided into “maternal” and “fetal”. Mothers at increased risk of congenital heart disease include those with

- a history of CHD in a first degree relative
- maternal diabetes
- maternal exposure to known cardiac teratogens

Findings arising in an individual pregnancy which increase the risk of CHD for that fetus include

- increased nuchal translucency
- the development of fetal hydrops
- the detection of any extracardiac malformation
- the detection of an arrhythmia
- the suspicion of CHD during an obstetric scan

“Maternal” Risk Factors

When a previous child has had congenital heart disease, the risk is increased in a subsequent pregnancy to 2%, with two previously affected pregnancies bringing the risk to 10%. Some malformations are more likely to be associated with recurrence than others, for example, left heart lesions and the heterotaxy syndromes. Recurrence is most commonly of the same lesion, but related or entirely different lesions can occur also. Thus, it is essential to know the nature of a previous malformation when a fetal echocardiogram is being performed, as it or related defects should be specifically looked for, in addition to excluding other forms of CHD.

Genetic defects, such as a microdeletion of chromosome 22 are increasingly recognised as being associated with specific types of congenital heart disease, such as common arterial trunk and interrupted aortic arch, and this sometimes leads to an unexpected diagnosis of a syndrome such as the velo-cardio-facial syndrome in a parent.

The difficulty in this setting is in predicting the risk of a cardiac malformation in future pregnancies. The risk of transmitting a microdeletion when a parent is affected is one in two, but the phenotype is very variable in this condition, perhaps due to the modifying influence of other genes. With the exception of this and other single gene defects, when a parent has congenital heart disease, the risk of recurrence usually lies between 5-10%. It may be higher when the mother is affected than the father although this is controversial. A mother with uncorrected cyanotic congenital heart disease has a low chance of conceiving and is at increased chance of spontaneous fetal loss. If the mother has palliated or repaired CHD, the risk will vary with the diagnosis and clinical status. If the family history of CHD is more distant than a first degree relative, the risk of CHD is probably the same as the normal population.

Maternal diabetes increases the risk of congenital heart disease by 3-5 times overall but there is a higher risk with poor control in early pregnancy and in association with high hemoglobin A1c levels.

Maternal exposure to either drugs or viral infection can damage the developing heart. Exposure to teratogenic drugs in the first 6-8 weeks of pregnancy increases the risk of CHD. These include alcohol, anti-convulsants, such as valproic acid and epanutin, retinoic acid and perhaps lithium. Recent evidence has challenged the popular belief that lithium is an important cardiac teratogen, which particularly caused Ebstein's malformation. The most common cardiac lesion seen after exposure to any of the other drugs is a ventricular septal defect but more complex lesions can occur. Maternal infection with parvovirus and coxsackie virus can cause a dilated cardiomyopathy in the fetus.

“Fetal” Risk Factors

Fetal hydrops is the development of fluid collections in more than one site, either in the abdomen (ascites), the pleural or pericardial space, or the skin. About 25% of cases of fetal hydrops examined by us have been found to have a cardiac etiology. The majority of cases are due to arrhythmias, either heart block or a tachycardia, but structural heart disease also occurs. In some cases, the cause of the hydrops may be an associated

chromosomal anomaly, but cases of left isomerism have a particular tendency to develop hydrops, especially if there is complete heart block.

When an extracardiac anomaly is found, the heart should be examined carefully. Malformations in more than one system may indicate a syndrome or chromosomal anomaly. Abnormalities which have particular association with CHD include

- Omphalocele
- Diaphragmatic hernia
- Duodenal atresia
- Nuchal edema
- Single umbilical artery
- Tracheo-esophageal fistula
- Cystic hygroma

However, complete cardiac evaluation is necessary after the detection of any extracardiac malformation, especially if the pregnancy is continuing. The association of omphalocele with CHD, which occurs in up to 30% of cases, will often indicate a chromosomal anomaly, but even if the chromosomes are normal, a cardiac malformation may influence the outcome of the surgical repair. The most commonly associated cardiac malformations in our series were tetralogy of Fallot, ventricular septal defect and atrioventricular septal defect. The incidence of CHD in diaphragmatic hernia may be as high as 10%. The same lesions as are seen with omphalocele are the most common. Duodenal atresia is commonly associated with an atrioventricular septal defect in the setting of trisomy 21, but ventricular septal defects are common when the chromosomes are normal. Nuchal translucency is thickening of the fluid space at the back of the neck. An increase in this space, which should measure less than 2.7mm at 10-12 weeks gestation, has a high association with Down's syndrome and other chromosomal anomalies. In those fetuses with an abnormal measurement and normal chromosomes, a high rate of CHD is found, with more severe disease related to an increasing measurement. The associated types of CHD are varied. Tracheoesophageal fistula often presents with hydramnios in the last ten weeks of pregnancy. It may be difficult to diagnose prenatally. Cardiac malformation in association is often part of a VATER syndrome and usually takes the form of tetralogy of

Fallot or ventricular septal defect. Cystic hygroma differs from nuchal edema in that it is a fluid space at the back of the neck which is septated. Typically it is associated with Turner's syndrome and coarctation of the aorta. In those cases with normal chromosomes, the fetal heart should be scanned for evidence of biventricular hypertrophy or pulmonary stenosis, which may lead to a diagnosis of Noonan's syndrome.

The detection of an abnormal cardiac rhythm should lead to a referral for fetal echocardiography. All cases of sustained bradycardia and all cases of tachycardia should have detailed cardiological assessment. An irregular rhythm is very common during pregnancy especially in the last ten weeks of gestation. These are due to ectopic atrial or ventricular beats. In the absence of structural malformation, these are usually of no significance.

Although it is important to offer fetal echocardiography to high risk patients, most congenital heart disease is found in otherwise normal, low-risk pregnancies. Congenital heart disease is common and is a cause of major mortality and morbidity in children. Thus, the concept of screening the normal obstetric population was introduced. Since screening the heart in a simplified fashion using the four-chamber view was first suggested by a French group in 1985, evaluation of the cardiac structure during obstetric scanning has become widespread and recommended by governing ultrasound bodies as the standard of practice. The four-chamber view will be abnormal in about 1 in 500 pregnancies and in about 60% of major CHD. A regional teaching program focusing on this view improved the detection rate of heart disease to about 70% of expected and this has continued to improve in subsequent years. However, overall, the detection rate of four-chamber view anomalies during routine scanning is currently running at about 50% in practice, with geographical variations within and between different countries. More recently, great artery evaluation has been added to the cardiac assessment during obstetric scanning to try to improve the detection rate of major CHD. Correct evaluation of both the four chamber and the great artery views will exclude about 90% of major cardiac malformations and has been successfully added to obstetric screening programs to yield a high rate of detection of CHD in some centers.