

Fetal Diagnostics and Fetal Intervention



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KEYWORDS

- Fetal echocardiography • Congenital heart disease • Fetal diagnostics
- Fetal intervention • Sequential segmental analysis

KEY POINTS

- Expectations for the accurate diagnosis of congenital heart disease in fetuses have increased significantly.
- Improved ultrasound technology allows for detailed anatomic and physiologic assessment of the fetal heart.
- Consistent application of a sequential segmental approach to the fetal echocardiogram ensures an accurate and comprehensive diagnosis to allow for appropriate perinatal management.



Video content accompanies this article at <http://www.perinatology.theclinics.com/>

INTRODUCTION

Congenital heart disease (CHD) is the most common type of birth defect; however, it is one of the most frequently missed abnormalities by prenatal ultrasound.¹ With advances in the fetal diagnosis of CHD and the availability of fetal therapies, it has become crucial to improve the screening of obstetric patients and correctly diagnose fetuses with CHD in utero. The fetal diagnosis of CHD impacts the prenatal and postnatal care of mothers and fetuses. Although the benefits of fetal diagnosis have been controversial, recent studies suggest that an accurate fetal diagnosis and actions taken as a result of a fetal diagnosis may improve morbidity and mortality, specifically for fetuses with critical CHD.^{2–5}

Disclosures: None.

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RATES OF FETAL DIAGNOSIS

The reported rates of fetal diagnosis of CHD vary greatly among regions and institutions. A recent study found that in the United States, the prenatal diagnosis rate of CHD was 26% in 2006 and increased to 42% in 2012.⁶ These data are similar to previously reported data from other centers.^{7,8} The rate of fetal diagnosis of CHD is affected by the type of cardiac lesion, ultrasound practice, and training of the examiner.^{7,8} Other factors, however, have been postulated to affect the fetal diagnosis of CHD, including maternal body habitus, maternal socioeconomic status, ethnicity, and geographic region of care.

OBSTETRIC SCREENING

Prenatal diagnosis of CHD requires a multidisciplinary approach. It requires access to timely prenatal care, efficient screening by trained sonographers, reliable interpretation, and the ability to refer to a pediatric cardiologist. A mother receiving prenatal care in the United States will almost universally receive a routine obstetric ultrasound.⁷ Despite having universal obstetric screening, rates of prenatal diagnosis of CHD vary greatly and remain low in certain areas.

The current guidelines from the American Institute of Ultrasound in Medicine, the American College of Radiology, and the American College of Obstetrics and Gynecology recommend that women should have a detailed anatomy screening ultrasound between 18 and 20 weeks of estimated gestational age, unless there is an indication for one earlier.⁹ The cardiac examination was initially limited to a four-chamber view of the heart. More recently, it has also included evaluation of the outflow tracts. Studies have shown that the four-chamber view can detect greater than 50% of serious cardiac malformations, but additional evaluation of the outflow tracts, including the “three-vessel” view, increases detection rates to 90%.¹⁰ This suggests that by following current guidelines, along with a more detailed assessment of the heart in a systematic fashion, an institution can significantly improve their rates of prenatal diagnosis of CHD.

FETAL ECHOCARDIOGRAPHY

Fetal echocardiography provides a detailed assessment of the structure and function of the fetal cardiovascular system to ensure an accurate diagnosis and guide prenatal counseling and perinatal management. Although obstetric screening has expanded its evaluation of the fetal heart, the fetal echocardiogram provides a dedicated fetal cardiac examination to include all cardiac structures and can further focus on the counseling regarding the pathophysiology of the cardiac lesion and future treatment options.

Indications for Fetal Echocardiography

Indications for fetal echocardiography are divided into maternal and fetal factors (**Box 1**). Recently, the American Heart Association published a complete list of indications for fetal echocardiography.¹¹ The leading indication for referral for a fetal echocardiogram is an abnormal obstetric screening ultrasound.¹² Conversely, of those fetuses with CHD who had a fetal echocardiogram, only 10% of those patients were referred for an identifiable risk factor.¹³ Therefore, all fetuses should at least undergo a comprehensive screening ultrasound by a trained professional.

Timing of Fetal Echocardiogram

The timing of the fetal echocardiogram should be determined by the reason for referral. Although fetal echocardiography can be performed in the first trimester, the optimal

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