

Fetal Ecocardiography as Screening Ultrasound for Every Pregnant Woman

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Editorial

The congenital heart defects are the most frequent malformations: their incidence (that is the number of new cases in a population), calculated as the number of children with heart disease on the total number of live births, is around eight cases per thousand live births; in Italy, approximately 4,000 infants every year are born with a congenital heart disease.

The incidence of congenital heart defects (CHD) has been fairly stable over the last 50 years, considering various international reports.

Moreover, CHD are the leading cause of infant mortality, with an estimated incidence of about 4-13 per 1000 live births [1-3]. The World Health Organization (WHO) recognized in the presence of congenital heart defects the cause of 42% of child deaths recorded between 1959 and 1994 [4].

Furthermore, structural abnormalities of the heart are some of the most frequent malformations whose diagnosis eludes the prenatal ultrasound evaluation [5-6].

Studies have shown that parents (especially mothers) affected by a congenital heart defect have a higher incidence of fetal heart defects.

It has been shown that prenatally recognition of CHD might improve the perinatal outcome of fetuses with specific types of cardiac lesions [7-12].

So, it may be reasonable to expect a slow increase in the incidence of congenital anomalies of the heart over the years, thanks to the newest medical and surgical therapies that help to heal children with these defects, giving them the opportunity to grow and, in turn, to have their own children.

The heart defect may be of various types, ranging from abnormalities that affect a single part of the heart (e.g., a valve) to very complex abnormalities, characterized by major changes in cardiac architecture.

The CHD severity and its effects on health are obviously different: some forms allow a normal life, while others are incompatible with life.

Having a congenital heart defect does not necessarily mean having a serious illness; however, some congenital heart defects are so complex that lead to death a newborn in a few days, unless it is made an immediate medical or surgical intervention.

The pioneering work of Kratochwil and Sassi in the '60s and the Kleimann's one in the 80s, up to the actual day, have always shown that fetal echocardiography is the only diagnostic tool for highlighting congenital heart disease [13-14].

The proper period for ultrasound evaluation of the fetal heart is the 22 weeks period of amenorrhea. Some abnormalities can be identified by the end of the first trimester or early second trimester, especially if it has been noticed an increased nuchal translucency [15-20].

However, many anatomical structures may continue to be satisfactorily analyzed even after 22 weeks, especially from a functional point of view.

For this reason, it is reasonable to do first the basic fetal cardiac ultrasound during the second trimester anomaly scan, and, if a major CHD is not suspected, it is good practice to suggest a fetal echocardiography screening at 26 weeks of pregnancy.

Moreover, Sumpfen's study showed that fetal echocardiography significantly improves the diagnostic capability of congenital heart disease verifying how the largest part of the heart diseases arises in the low-risk population. This evidence was confirmed by Yagel too. It is well known that the 4-heart chambers view allows the diagnosis of only 50% of cardiac anomalies. The visualization of the 4-heart chambers and cardiac outflows is part of the routine second trimester ultrasound scan, with a sensitivity of 78%-83%. However, when this scan is performed in peripheral centers there is a failure of 65%-90% in the diagnosis of heart disease. Screening methods that have the aim of identify fetuses who need a detailed study of cardiac anatomy (as the nuchal translucency > 2.5 mm) have failed. Failure of prenatal diagnosis of congenital heart disease increases the psychological distress of the couple (with relative increase in litigation coroner) and the management costs for an infant with complications related to the failed diagnosis.

From these data it is evident that the study of the fetal heart during the anomaly scan have failed in the early diagnosis, suggesting how important is to propose screening of such anomalies through an echocardiographic study offered to all pregnant women regardless of risk of recurrence of discrepancy in object.

Indeed, the pediatric cardiologist will be the appropriate figure to perform an adequate counselling, discussing with the couple about intrauterine prognosis and intrauterine treatment, planning the prenatal and post-natal follow-up, according with the timing of the delivery.

In the Netherlands a national prenatal heart screening programme was introduced in 2007. A very recent study evaluates the effects of this screening programme. It was the largest cohort study to investigate the prenatal detection rate of severe CHD in an unselected population. Cases were divided into two groups: before and after the introduction of screening. A nationally organised screening has resulted in a remarkably high detection rate of CHD (59.7%) compared with earlier literature [21].

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Received: December 07, 2015; **Accepted:** December 09, 2015; **Published:** December 16, 2015

Citation: Stracquadano M, Fauzia M, Palumbo M, Emidio LD, Giorlandino C, et al. (2015) Fetal Ecocardiography as Screening Ultrasound for Every Pregnant Woman. J Preg Child Health 2: e127. doi:[10.4172/2376-127X.1000e127](https://doi.org/10.4172/2376-127X.1000e127)

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