False positives after the ultrasound evaluation of the fetal heart at the time of first trimester scan: an Italian multicentric study

SIEOG Working Group of Regional Coordinators

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Introduction

When carried out by experienced operators, first trimester assessment of the fetal heart can lead to the early detection of major congenital heart disease. However, false positive cases may be responsible for increased level of anxiety of the parents and unnecessary medical interventions.

Objectives

To evaluate the false positive results following fetal cardiac anatomical ultrasound assessment in the first trimester in a low risk population.

Methods

This was an Italian multicentric retrospective study of pregnant women undergoing first trimester combined screening for aneuploidies in six fetal medicine units. In all cases an ultrasound evaluation of the fetal heart was performed by obstetricians with extensive experience in first trimester scan. The protocol for the assessment of the fetal heart included: an axial view of the upper abdomen which, together with levocardia, helps determining fetal visceral situs, the four chamber view and the three vessels and trachea view. All these views were obtained by grayscale ultrasound and Colour flow mapping. In all suspected abnormal cases a fetal echocardiography was offered in later stages of pregnancy.

Results

Among the 4.300 fetuses included in the study, forty-six congenital heart defects were suspected between 11 and 13+6 weeks of gestation. Twenty-four cases were excluded from the analysis because parents opted for early termination of pregnancy due to associated anomalies, including abnormal fetal karyotype and/or other fetal structural abnormalities. Postmortem evaluations were not available. For the remaining 22 fetuses echocardiography was performed by the 16th week of gestation in 14 cases (64%) and after 16 weeks in 8 cases. In 19 cases (86%) the fetal cardiologist confirmed the presence of a congenital heart defect and in 16 out of these 19 cases parents opted for termination of pregnancy. In 3 cases the cardiac anatomy resulted normal at the fetal echocardiography. An amniocentesis was performed in 2 out of these 3cases. The indication for invasive procedure was an increased risk for aneuploidies at the first trimester screening in one case, and the suspect of the fetal heart defect in the other. In both cases fetal karyotype was normal and Chromosomal Microarray Analysis did not detect clinically significant copy number variations. Postnatal echocardiography was normal for these 2 cases, while the third pregnancy is still ongoing.

Maternal Characteristics	Value
Maternal age (years)	35 (25-47)
Body Mass Index (Kg/m2)	24 (17-40)
Gestation age (weeks)	12+3 (11+0- 14+0)
Pregnancy	
Singleton	44 (96%)
Multiple	2 (4%)
Mode of conception	
Spontaneous	41 (89%)
IVF	5 (11%)
Smoking	4 (9%)

Fetal characteristics	Value
Nuchal translucency thickness (mm)	3.35mm (1.1-13.6)
Nuchal translucency thickness >99th	29 (50%)
Nuchal translucency thickness >95th	32 (69%)
First trimester increased risk for aneuploidies	29 (63%)
Abnormal 4 chamber view	19 (41%)
Abnormal 4 chamber view and 3 vessel trachea view	23 (50%)
Abormal 3 vessel trachea view	4 (9%)
Associated fetal abnormalities	25 (54%)
Invasive procedures (CVS or Amnio)	36 (78%)
Genetic or chromosomal anomalies	22 (64%)

Conclusions

Ultrasound evaluation of the fetal heart at the time of first trimester scan performed by experienced operators allowed to anticipate the diagnosis of fetal heart defects performing a fetalechocardiography within 16 weeks of gestation in most cases, giving more time for additional investigations and informed parental decision making around the course of pregnancy. The false positive rate at the first trimester ultrasound examination of the fetal heart has been low. However, it should be kept in mind that false positives can increase parents anxiety and over-medicalization of the pregnancy. During counselling, the parents should be informed that an ultrasound abnormal finding at the first trimester examination of the fetal heart does not mean that acongenital heart disease is present. This is the reason why invasive procedures should be performedonly when the suspects are confirmed at the fetal echocardiography or if other indications emerge at the first trimester scan.