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REVIEW

Ultrasonographic soft markers of aneuploidy in second trimester fetuses

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KEYWORDS

Ultrasound;
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Aneuploidy;
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Abstract *Objective:* To evaluate ultrasound "soft markers" used in fetal genetic screening.

Options: Ultrasound screening at 16–20 weeks is one of the most common genetic screening tests used during pregnancy. The practical concern for ultrasound screening is false-positive and false-negative results. The use and understanding of ultrasound soft markers and their screening relative risks are an important option in the care of pregnant women.

Introduction: Chromosomal abnormalities occur in 0.1–0.2% of live births, and the most common clinically significant aneuploidy among live-born infants is Down's syndrome (trisomy 21). Soft markers of aneuploidy are nonspecific, often transient, and can be readily detected during the second and third trimester ultrasound. The most commonly studied soft markers of aneuploidy include a thickened nuchal fold, mild fetal pyelectasis, echogenic bowel, echogenic intracardiac focus and choroid plexus cyst. There is a great deal of interest in the ultrasound detection of aneuploidy, as evidenced by the large number of publications in the literature on this topic.

Contents

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1. Introduction

Chromosomal abnormalities occur in 0.1–0.2% of live births (1,2). Trisomy 21 (Down's syndrome) is the most common chromosomal abnormality in live-born infants (1 per 800 live births) (3). Sonographic findings in fetuses with Down's syndrome include both structural and nonstructural markers. However, other aneuploidies like trisomy 13, trisomy 18, monosomy X, and triploidy can also be detected by ultrasound.

Many methods have been used to identify women at risk of carrying a fetus with aneuploidy, including maternal age (4), biochemical markers (5), prenatal ultrasound and amniocentesis (6,7), However, there is a 0.5–1.0% fetal mortality associated with this invasive procedure (8,9).

A second-trimester ultrasound scan is usually done at 16–20 weeks. Two types of sonographic markers suggestive of aneuploidy can be observed in the second trimester. Major fetal structural abnormalities and soft markers of aneuploidy are less-defined, less significant and often transient (Table 1). Although these markers are not pathognomonic because they may be seen in the normal fetus but they have been used to screen for Down's syndrome and other aneuploidies (10,11). Thus, prenatal ultrasonography during the second trimester provides a ''genetic sonogram'' that is used to identify structural features of fetal Down's syndrome (12,13).

The most commonly studied soft markers of aneuploidy include a thickened nuchal fold, limb shortening, mild fetal pyelectasis, echogenic bowel, echogenic intracardiac focus (EIF), choroid plexus cyst (CPC) and single umbilical artery. We review the most common ultrasonographic soft markers used to screen aneuploidy and discuss ultrasonographic technique and measurement criteria for the detection of soft markers. We also review the clinical assessment of soft markers for aneuploidy risk.

2. Sonographic soft markers of aneuploidy

2.1. Thickened nuchal fold (Fig. 1)

2.1.1. Definition and imaging criteria

The nuchal fold is the skin thickness in the posterior aspect of the fetal neck. A nuchal fold measurement is obtained in a transverse section of the fetal head at the level of the cavum septum pellucidum and thalami directed posteriorly to the cerebellum (14). The measurement is taken from the outer edge of the occiput to the outer skin in the midline. The definition of a thickened nuchal fold is that a measurement 6 mm or more between 15 and 23 weeks. However, a thickened nuchal fold should be distinguished from cystic hygroma, in which the skin in this area has fluid-filled Cysts. A thickened nuchal fold differs from nuchal translucency, which is a specific measurement