Evolution of Counseling for Isolated Soft Ultrasound Markers

TITLE: Ultrasound Isolated Soft Markers

Subtitle: Evolution of Counseling

SUMMARY: Soft ultrasound (US) markers were introduced to improve the detection of Trisomy 21 (T21) over age-related risk alone. The most commonly reported soft markers include echogenic intracardiac focus (EIF), echogenic bowel, choroid plexus cyst (CPC), single umbilical artery (SUA), urinary tract dilation (UTD), Shortened humerus/femur/or both, thickened nuchal fold (NF), absent/hypoplastic nasal bone (AHNB). These minor US findings do not represent structural abnormalities and are often normal variants, but demonstrate association with an increased aneuploidy risk. cfDNA is the single best screening test for common trisomies and results in the lowest residual risk of aneuploidy. When an isolated soft marker is identified after negative screening, patients can be reassured that the risks of fetal aneuploidy remain low. Only diagnostic testing removes residual risk.

Rationale: The management recommendations for isolated soft markers (ie no structural anomalies, fetal growth restriction, or additional markers) have evolved in the current era of widespread cell-free DNA (cfDNA) screening with its high sensitivity and specificity for T21, 18, 13 across all age groups. Because most soft markers have low likelihood ratios for a particular aneuploidy, the post-test probability, after a low risk cfDNA result, is very low and diagnostic testing (amniocentesis, chorionic villus sampling) is no longer recommended. In the case of normal results on alternative serum screening (first trimester screen, integrated/sequential/contingent screen, quad screen), identification of most soft markers also results in low post-test probability and additional screening/testing is not recommended. The exceptions are isolated thickened NT and absent/hypoplastic NB for which additional screening by cfDNA and diagnostic testing should be discussed.

Eligible patients: All pregnant patients with an intrauterine pregnancy and isolated soft marker on ultrasound

Contraindications: Nonviable pregnancy

Technique: All patients should be offered screening (serum +/- nuchal translucency based on local standards) and diagnostic testing (chorionic villus sampling, amniocentesis) early in pregnancy as a matter of routine prenatal care. All patients with an apparent isolated soft marker should be offered a detailed US (CPT 76811, targeted scan). Not all patients will desire such screening, invasive testing, or detailed US.

Special Considerations:

1. In the setting of a normal karyotype on diagnostic testing, isolated soft markers do not present a concern for aneuploidy although other considerations may persist (eg- echogenic bowel and risk for CF/CMV).
2. Patients with normal serum screening and isolated thickened NT or absent/hypoplastic NB should consider additional screening by cfDNA or diagnostic testing due to the higher risk for aneuploidy associated with these markers.

Reference: SMFM Consult Series #57, AJOG 2021 OCT; 225(4)B2-B15.

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