Environmental risk factors and hepatic disorders during pregnancy.

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Introduction

Soft markers were originally introduced to prenatal ultrasonography to improve the detection of trisomy 21 over that achievable with age-based and serum screening strategies. As prenatal genetic screening strategies have greatly evolved in the last 2 decades, the relative importance of soft markers has shifted. The purpose of this document is to discuss the recommended evaluation and management of isolated soft markers in the context of current maternal serum screening and cell-free DNA screening options. In this document, "isolated" is used to describe a soft marker that has been identified in the absence of any fetal structural anomaly, growth restriction, or additional soft marker following a detailed obstetrical ultrasound examination. In this document, "serum screening methods" refers to all maternal screening strategies, including first-trimester screen, integrated screen, sequential screen, contingent screen, or quad screen [1].

The Society for Maternal-Fetal Medicine recommends the following approach to the evaluation and management of isolated soft markers: we do not recommend diagnostic testing for aneuploidy solely for the evaluation of an isolated soft marker following a negative serum or cell-free DNA screening result (GRADE 1B); for pregnant people with no previous aneuploidy screening and isolated echogenic intracardiac focus, echogenic bowel, urinary tract dilation, or shortened humerus, femur, or both, we recommend counselling to estimate the probability of trisomy 21 and a discussion of options for non-invasive aneuploidy screening with cell-free DNA or quad screen if cell-free DNA is unavailable or cost-prohibitive (GRADE 1B);for pregnant people with no previous aneuploidy screening and isolated thickened nuchal fold or isolated absent or hypo plastic nasal bone, we recommend counselling to estimate the probability of trisomy 21 and a discussion of options for noninvasive aneuploidy screening through cell-free DNA or quad screen if cell-free DNA is unavailable or cost-prohibitive or diagnostic testing via amniocentesis, depending on clinical circumstances and patient preference (GRADE 1B); for pregnant people with no previous aneuploidy screening and isolated choroid plexus cysts, we recommend counselling to estimate the probability of trisomy 18 and a discussion of options for noninvasive aneuploidy screening with cell-free DNA or quad screen if cell-free DNA is unavailable or cost-prohibitive [2-3].

For pregnant people with negative serum or cell-free DNA screening results and an isolated echogenic intracardiac focus, we recommend no further evaluation as this finding is a normal variant of no clinical importance with no indication for fetal echocardiography, follow-up ultrasound imaging, or postnatal evaluation. For pregnant people with negative serum or cell-free DNA screening results and isolated fetal echogenic bowel, urinary tract dilation, or shortened humerus, femur, or both, we recommend no further aneuploidy evaluation (GRADE 1B); for pregnant people with negative serum screening results and isolated thickened nuchal fold or absent or hypo plastic nasal bone, we recommend counselling to estimate the probability of trisomy 21 and discussion of options for no further aneuploidy evaluation, non-invasive aneuploidy screening through cell-free DNA, or diagnostic testing via amniocentesis, depending on clinical circumstances and patient preference (GRADE 1B); for pregnant people with negative cell-free DNA screening results and isolated thickened nuchal fold or absent or hypo plastic nasal bone [4]. We recommend no further aneuploidy evaluation (GRADE 1B); for pregnant people with negative serum or cell-free DNA screening results and isolated choroid plexus cysts, we recommend no further aneuploidy evaluation, as this finding is a normal variant of no clinical importance with no indication for follow-up ultrasound imaging or postnatal evaluation (GRADE 1C); for fetuses with isolated echogenic bowel, we recommend an evaluation for cystic fibrosis and fetal cytomegalovirus infection and a third-trimester ultrasound examination for reassessment and evaluation of growth (GRADE 1C); for fetuses with an isolated single umbilical artery, we recommend no additional evaluation for aneuploidy, regardless of whether results of previous aneuploidy screening were low risk or testing was declined. We recommend a third-trimester ultrasound examination to evaluate growth and consideration of weekly antenatal fetal surveillance beginning at weeks of gestation (GRADE 1C); for fetuses with isolated urinary tract dilation A1, we recommend an ultrasound examination at ≥ 32 weeks of gestation to determine if postnatal paediatric urology or nephrology follow-up is needed. For fetuses with urinary tract dilation [5]. We recommend an individualized follow-up ultrasound assessment with planned postnatal follow-up (GRADE 1C); for fetuses with isolated shortened humerus, femur, or both, we recommend a third-trimester ultrasound examination for reassessment and evaluation of growth.

References

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