SIGNIFICANCE OF SONOGRAPHIC SOFT MARKERS OF FETAL ANEUPLOIDY IN DAY TO DAY PRACTICE

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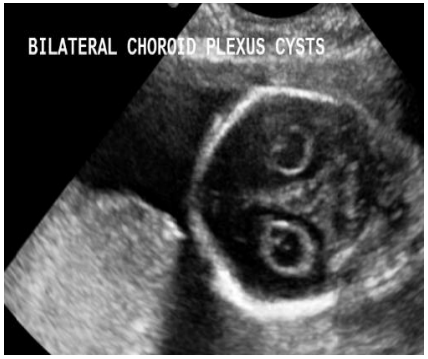
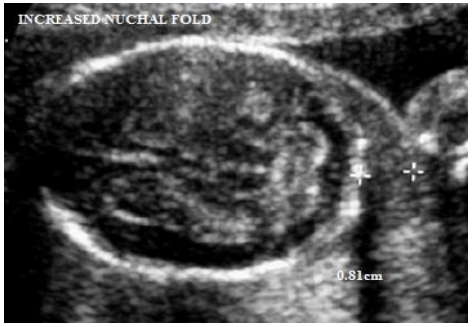
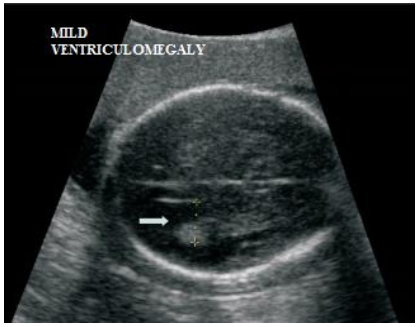
INTRODUCTION

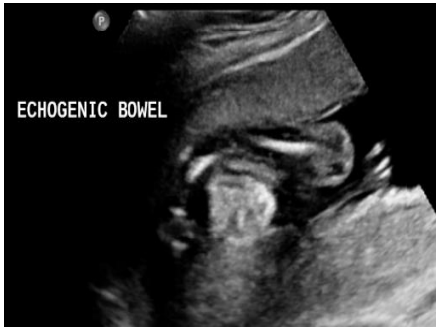
Soft markers are minor ultrasound findings readily detected during a genetic sonogram, considered variants of normal, and do not constitute a structural defect. They are non-specific and often transient and may be associated with chromosomal or non chromosomal abnormalities. The presence of soft markers increases the risk of fetal aneuploidy but is not diagnostic. Individual soft marker will vary in degree of association with fetal aneuploidy.

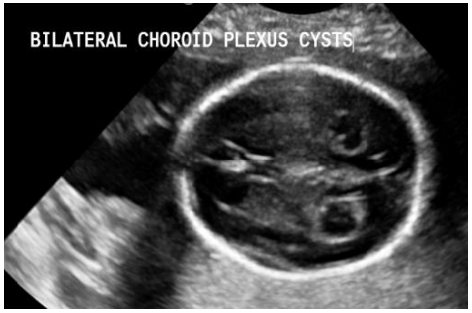
DISCUSSION About sixteen potential second trimester soft markers can be identified. Only 5 markers are considered useful for evaluation of fetal aneuploidy which include increased nuchal fold thickness, echogenic bowel, mild ventriculomegaly, echogenic focus in the heart and choroid plexus cyst. Although not pathologic of themselves, these soft markers have been used to screen for or adjust the risk for Down's syndrome and other aneuplodies. It has become practice to estimate the degree of association as likelihood ratio (LR) by which the priori background risk is altered. Detection of multiple soft markers will increase the significance of findings, compared with screening the same marker in isolation. Non sonographic factors including maternal age, gestational age, past history and family history also influence the chance of aneuploidy and should be considered to establish an accurate a priori risk.

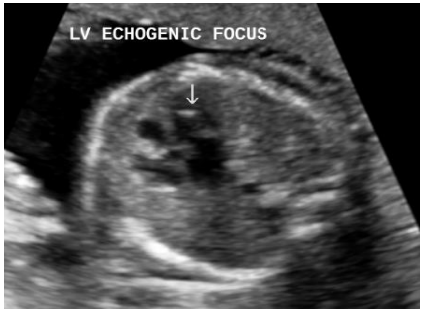


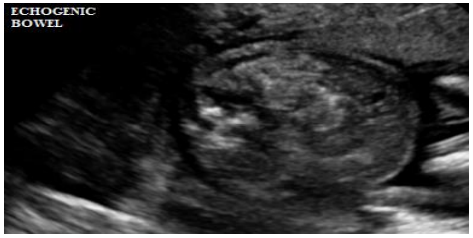


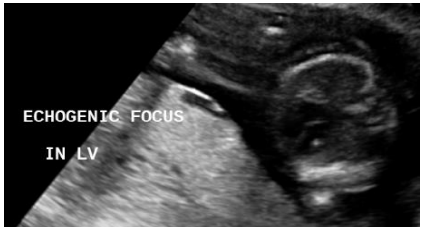












The detection of any abnormal finding on ultrasound should prompt an immediate detailed ultrasound evaluation of the fetus by an experienced sonographer. Referral to a tertiary center and karyotyping should be considered if there are more than one abnormal ultrasound finding, if the patient is over 35 years of age or if the bio chemical screening is abnormal. Traditionally , amniocentesis is generally warranted when the risk is of 1 in 270 or greater.

CONCLUSION:

The bottom line is that, in an otherwise low risk pregnancy with a normal first trimester screening the presence of one soft marker does not significantly raise the odds ratio and more invasive testing is not usually warranted. As early screening (nuchal translucency, early maternal serum testing) and diagnosis (CVS) become established, the significance of second trimester markers will decrease and require readjustment.

References:
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