

Clinical significance of fetal nuchal translucency during early pregnancy

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● Objective

To examine the significance of fetal nuchal translucency during early pregnancy in detection of abnormal karyotype and major cardiac defect.

● Material & Methods

Between Jul 1998 and Jul 2002, ultrasound examination was performed in 1253 unselected singleton pregnancies with a live fetus and fetal crown rump length of 24-88 mm. The fetal nuchal translucency thickness was measured successfully in all cases. We reviewed the pregnancy outcome about two parameters. Abnormal karyotype was confirmed by antenatal karyotyping and newborn's feature suggestive of a chromosomal defect. Major cardiac defect was confirmed by newborn physical examination, targeted ultrasonography, neonatal echocardiography and autopsy.

● Results

(1) Fetal nuchal translucency thickness increased significantly with crown-rump length. (2) Nuchal translucency was greater than 3 mm in 12 pregnancies with abnormal karyotypes, giving a detection rate of 75.0% with a false positive rate of 4.0%. With a cutoff of 2.5 mm, a detection rate of abnormal karyotype was 81.3% with a false positive rate of 8.3%. (3) With normal karyotype, the overall prevalence of major cardiac defects in this study population was 5/1000 pregnancies (5/1054). This prevalence increased from 48/1000 (4/83) using 2.5 mm cutoff to 105/1000 (4/38) using 3 mm cutoff. The detection rate for major cardiac defect of nuchal translucency above 3 mm is 80.0% with a false positive rate of 3.2 percent.

● Conclusion

Increased nuchal translucency is by far the single most efficient marker for screening of chromosomal defects and most effective screening method for major cardiac defects.