

Risk for isolated ventricular septal defect in euploid fetuses with borderline or increased first trimester nuchal translucency.

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INTRODUCTION

The association of an increased nuchal translucency (NT) in the first trimester with a higher prevalence of fetal complex structural cardiac defects, in the absence of chromosomal abnormalities, has long been reported. Its association with isolated interventricular septal defect is unknown.

OBJECTIVE

To test the hypothesis that there is association between a borderline or increased NT (2 mm or more) in 11+0 - 13+6 weeks euploid fetuses and a later diagnosis of isolated VSD.



METHODS

A case-control study was designed. 5464 second or third-trimester consecutive fetuses without extracardiac abnormalities and no cardiac anomalies other than a VSD were assessed in a period of two years. A fetal echocardiogram and a morphological scan were performed, searching for the diagnosis of VSD and to exclude associated cardiac and noncardiac malformations. Fetuses with an altered karyotype or a postnatal diagnosis of chromosome abnormalities were excluded. Statistical analysis used Fisher's exact test and ROC curves.

RESULTS

TN < 2

5180 FETUSES

115 CIV (2.2%)

(86 muscular, 29 perimembranous)

TOTAL

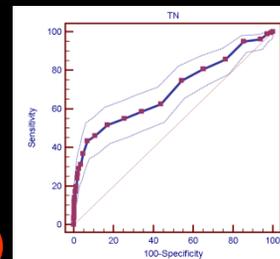
5464 FETUSES

TN ≥ 2

319 FETUSES

67 CIV (21%)

(52 muscular, 15 perimembranous)



A ROC curve determined the cut-off value of NT (2.0), with a sensitivity of 48.3% and a specificity of 91.4%, with an area under the curve = 0.695 (p < 0.0001).

Significant association (p < 0.0005)
RR = 9.3 (99% CI : 6.5 – 13.5)

CONCLUSIONS

Fetuses without chromosome abnormalities with a first trimester NT of 2mm or more have an 8.3-fold increase in the risk of presenting an isolated ventricular septal defect. We speculate that the defects could have been larger and functionally important in the first trimester, increasing the NT as a consequence of hemodynamic overload and gradually decreasing its diameter until the second and third trimesters. This knowledge may have implications in prenatal management and counseling.