

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

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Basics: 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. The purpose of this study was 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. NT had been obtained between 11+0 and 13+6 weeks of gestation. 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: Mean maternal age was 32±5 years (21-42 years) and gestational age at the time of the fetal echocardiogram was 25±6 weeks (19-31 weeks). Mean NT was 2.2 in fetuses with VSD and 1.4 without. Out of 319 fetuses with a NT of 2 mm or more, 67 had a VSD (52 muscular and 15 perimembranous) (21%), while in only 115 out of 5180 fetuses with a NT<2,0mm a VSD was detected (86 muscular and 29 perimembranous) (2.2%) ($p<0.0005$), being the relative risk = 9.3 (99% CI: 6.5-13.5). 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 ($p<0.0001$).

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.