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Increased nuchal translucency before 11 weeks of gestation

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Objective

An increased nuchal translucency (NT) ≥3.5mm is a reliable marker for chromosomal- and structural anomalies between 11 and 13^{+6} weeks gestation. Little is known about its performance as a screening tool before 11 weeks of gestation. Therefore, we wanted to investigate whether fetuses with an increased NT before 11 weeks of gestation and a normalized NT thereafter, are at risk for an adverse pregnancy outcome.

Methods

This is a prospective cohort study including pregnant women with a viable fetus with a crown-rump length (CRL) <45mm and a NT ≥2.5mm. All women were referred to our fetal medicine unit (FMU) one week later where the NT was remeasured with a CRL >45mm. Two groups were constructed: one in which the NT has normalized (<3.5mm) and one with a persistently increased NT (≥3.5mm). We monitored the cases until four weeks after delivery. The main outcome is a composite adverse outcome of chromosomal-, genetic, structural anomalies and pregnancy loss. We made subgroup analyses based on normalized or persistently increased NT at follow-up, and for NT thickness at inclusion.

Results

We included 110 cases of which 39 (35.5%) had an abnormal pregnancy outcome. Mean CRL and NT at inclusion and follow-up were 38.6 ±4.2mm and 3.9 ±1.1mm, and 52.7 ±4.8mm and 3.5 ±2.5mm, respectively. In the normalized NT group an abnormal outcome was found in 9/64(14%), of which 5 aneuploidies (7.8%). This was significantly different (p<.001, OR 11.5) compared to the persistently increased NT group, with an abnormal outcome in 30/46 (65%), of which 21 aneuploidies (46%). Subgroup analysis of NT 2.5-3.4mm at inclusion showed an abnormal outcome in 22%, and 16% if the NT normalized and was thus never ≥3.5mm.

Conclusion

Fetuses with an early increased NT thickness are at high risk of an abnormal pregnancy outcome, even if the NT normalizes after 11 weeks. Not all congenital anomalies could be found with routine first trimester screening. Therefore, these results showed that expectant parents should be referred to a FMU for detailed ultrasonography and invasive diagnostic should be offered, if an thickened NT is seen with “eye-balling” during a dating scan, even before 11 weeks.