NUCHAL TRANSLUCENCY AND CARDIAC DEFECTS

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Introduction
Nuchal translucency (NT) - a well described sonographic sign at 11-14 weeks of gestation is imaged as a sonotranslucent area behind the fetal neck. Normal NT thickness (measured in mm) is dependent on fetal size i.e. crown-rump length. Increased NT thickness - initially recognised as a sonographic marker for chromosomal defects\(^1\) has subsequently been shown also to be associated with major congenital heart disease (CHD). Whilst the initial reports pointed towards such an association in fetuses with aneuploidy,\(^2\)\(^-\)\(^4\) this has also been demonstrated in chromosomally normal fetuses.\(^7\) Data from nearly 1,500 fetuses with increased NT strongly suggested this to be a potentially useful marker for major cardiac defects and also suggested that the incidence of CHD increased with increasing NT measurements.\(^7\) Over the last decade there has been growing evidence to support increased NT at 11-14 weeks of gestation as a new marker for major CHD.\(^6\)\(^-\)\(^13\)

Increased NT – an indication for fetal echocardiography
The higher the NT measurement in a chromosomally normal fetus, the higher the risk of encountering a major heart abnormality. Much of the published data still refer to NT thickness of 2.5 mm as a cut-off point for the 95th centile as these studies reflect early information in this field even though it is now clear that reference ranges for the 95\(^{th}\) centile depend on fetal size. For the 99th centile however, a value of 3.5 mm can be taken as this centile limit for different fetal sizes.

The risk of a major cardiac abnormality in fetuses with NT ≥99th centile (i.e., NT ≥3.5 mm) appears to be around 5-10% (7;8;12;14). Thus, this is a clear indication for specialised fetal echocardiography. On the other hand, for NT measurements ≥2.5mm but <3.5 mm, the incidence seems to be marginally higher (17/1000)\(^14\) than that of the general population. This NT related risk (1.7%) is similar to other risk factors generally accepted as indications for detailed assessment of the fetal heart such as a previous child with CHD.\(^7\) Based on this, NT ≥2.5 mm also constitutes an indication for referral. However, if this cut-off point is to be used, approximately 5% of the population would have to be referred for fetal echocardiography. Available human and financial resources will limit the utility of using this cut-off point when the risk of major CHD is still very close to that of the general population.

Knowing the absolute value of NT provides further stratification of risks.\(^7\) For example, if NT lies between 3.5-4.4 mm the risk is around 3%, but increases further to ~ 10% and ~ 20% if NT ranges are 4.5-6.5 mm and 6.5-8.5 mm respectively.\(^16\) As most fetuses with CHD will still have normal NT measurements,\(^7\)\(^11\) this marker ought to be seen as an additional tool to help detecting CHD prenatally. It is still very important that at the time of the routine ‘anomaly scan’, all fetuses have a good obstetric screening using the the four-chamber view and possibly great vessel assessment as this remains the most important means of effectively diagnosing CHD before birth.\(^18\)
Following detection of an increased NT and exclusion of any associated chromosomal defect it is not possible to predict the type of structural cardiac abnormality to be found if there is one. Many different forms of CHD have been reported in association with previously increased NT with many reports showing there to be a wide spectrum of abnormalities.\textsuperscript{7,8,17,19-21} Knowing that NT was increased allows referral for specialist fetal echocardiography to be made earlier\textsuperscript{17} particularly if NT >4 mm.

**Increased NT and early fetal echocardiography**

One of the reported advantages of knowing the increased risk of having a child with major CHD following NT assessment at the time of the ‘11-14 week scan’ is the awareness that such risk exists early in the pregnancy. The feasibility of performing transabdominal\textsuperscript{22} as well as transvaginal echocardiography from the late first trimester together with realisation of NT-related risks have contributed enormously to the growing interest in performing fetal echocardiography at less than 16 weeks of gestation\textsuperscript{23,24-27} Referral for early cardiac assessment (from around 12-13 weeks), however, is not widely available. If performing an early fetal echo, it is important to realise that an important proportion of euploid fetuses with increased NT will have a normal heart and that a proportion of these will have significant extracardiac abnormalities. This stresses the importance also to consider early obstetric assessment if patients are referred for early fetal echocardiography.\textsuperscript{28}

**References**

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