

term “nuchal translucency” to describe the sonographic feature of normal fluid accumulation between the dorsal edge of soft tissue of the fetal neck and the skin, extending for variable distance over the fetal head and back, irrespective of whether it is septated or not<sup>6</sup> (Fig. 1). A significant correlation of NT thickness and risk for Down’s syndrome was demonstrated. An association between increased NT measurement and higher risk of other fetal aneuploidies, such as trisomy 18 and 13, Turner syndrome and triploidy was subsequently established.<sup>7</sup> Moreover, in the absence of chromosomal abnormalities, a higher risk of a variety of fetal structural, functional and genetic aberrations has been reported.<sup>8,9</sup> The Fetal Medicine Foundation (FMF) in England has established technical requirements for a standardized NT measurement which have been adopted by many other countries, especially in Europe (Table 1). In the United States, the Nuchal Translucency Quality Review (NTQR) program has recently specified corresponding guidelines (Table 2). The main difference concerns the optimal gestational age for NT measurement: FMF suggests 11 weeks to 13 weeks and 6 days or crown-rump length (CRL) 45 to 84 mm whereas NTQR recommends 10 weeks and 3 days to 11 weeks and 6 days or CRL 38 to 84 mm. FMF rationalizes the selection of 11 weeks as the lower limit since the safety of chorionic villus sampling (CVS) performed earlier, if indicated, is not acceptable. Furthermore, 11 weeks of gestation allow the detection of major fetal defects, such as acrania or exomphalos, which would otherwise be missed at NT scan. The screening process requires appropriately trained sonographers as well as regular audit of results and continuous assessment of image quality, in order to be reliable. It was estimated that using a risk cut-off for Down’s syndrome of 1 in 300, NT scan would identify approximately 80% of trisomy 21 fetuses for a false positive rate of 5%. The reported sensitivity of this procedure differs in various studies. In the largest one conducted so far, coordinated by the FMF, in 96,127 pregnancies followed after NT sonographic examination, the aforementioned estimated risk cut-off would identify 82% of trisomy 21 pregnancies and 78% of those with other aneuploidies for a false positive rate of 8%, equivalent to 77% detection rate of trisomy 21 for a 5% false positive rate.<sup>10</sup>

### The Issue of First Rather than Second-trimester Screening for Aneuploidies

Two main advantages of first rather than second-trimester screening for chromosomal abnormalities have been uniformly recognized so far. Firstly, early reassurance of fetal well being eliminates both maternal anxiety and uncertainty regarding the present gestation. Secondly, early diagnosis of an abnormal fetus allows decision making and potential subsequent termination of pregnancy in the first-trimester of pregnancy where complication rates are lower. Moreover, in terms of privacy,



Fig. 1: A standardized view of a fetus for nuchal translucency (NT) measurement

Table 1: Technical requirements for a standardized NT measurement proposed by FMF

1. Gestational age must be between 11 and 13<sup>+6</sup> weeks
2. The fetal crown-rump length should be between 45 and 84 mm
3. A good sagittal section of the fetus must be obtained, with the fetus horizontal on the screen. The correct view is a clearly visualised fetal profile
4. The fetus should be in a neutral position, with the head in line with the spine, not hyper-extended or flexed
5. Ideally only the fetal head and upper thorax should be included. The magnification should be as large as possible and always such that each slight movement of the calipers produces only a 0.1mm change in the measurement
6. The widest part of translucency must always be measured
7. Measurements should be taken with the inner border of the horizontal line of the calipers placed ON the line that defines the nuchal translucency thickness—the crossbar of the caliper should be such that it is hardly visible as it merges with the white line of the border, not in the nuchal fluid
8. In magnifying the image (pre or post freeze zoom) it is important to turn the gain down. This avoids the mistake of placing the caliper on the fuzzy edge of the line which causes an underestimate of the nuchal measurement. Do not use tissue harmonic imaging for measurement of nuchal translucency because this thickens the lines and underestimates the measurement
9. Care must be taken to distinguish between fetal skin and amnion

During the scan more than one measurement must be taken and the maximum one that meets all the above criteria should be recorded in the database. It is good practice to retain at least one image for your patient records.

NT, nuchal translucency; CRL, crown rump length

Adapted from FMF official Web site “<http://www.fetalmedicine.com/f-downs.htm>”

the earlier an abnormal pregnancy is terminated the lesser the chance of being widely recognized. On the other hand, a possible disadvantage of earlier screening is the detection and subsequent