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 not6 (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 aneuploides, such as trisomy 18 and 13, Turner syndrome and triploidy was subsequently established.7 Moreover, in the absence of chromosomal abnormalities, a higher risk of a variety of fetal structural, functional and genetic aberrations has been reported.8,9 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 aneuploides for a false positive rate of 8%, equivalent to 77% detection rate of trisomy 21 for a 5% false positive rate.10

The Issue of First Rather than Second-trimester Screening for Aneuploides

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, 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