Improved methodology for evaluating nuchal thickness in the second trimester

Nuchal thickening remains one of the most sensitive and important markers of Down syndrome in the second trimester of pregnancy and is universally used (Nyberg and Souter, 2001). In the second trimester the standard view for evaluating nuchal thickness is the modified transverse scan of the fetal head and neck (Benacerraf and Frigoletto, 1987). The measurement is made from the outer occipital bone to the outer skin line, and a nuchal thickening of 6 mm or greater is interpreted as increasing the risk for Down syndrome. There have been nine prospective studies addressing the validity of midtrimester nuchal thickness in the prediction of Down syndrome with sensitivities ranging from 16% to 75% (median of 39% and mean of 41%), with false-positive rates ranging from 0% to 8.5% (median of 0.85%, mean of 1.39%, and composite mean of 2.15%) (Borrell *et al*., 1996). There have been recent attempts to increase sensitivity by using a 5 mm cut-off; however, this has resulted in an increase of the false-positive rate from 0.1% to 2.1% (Borrell *et al*., 1996) and from 1% to 6.3% (Gray and Crane, 1994). In the paper with zero false-positive rate, nuchal thickening of 6 mm or more identified 41% of trisomy 21 fetuses and 50% of trisomy 18 fetuses (Ginsberg *et al*., 1990). This zero false-positive rate was achieved through the use of the dorsal midsagittal scan. When there was any question as to whether the thickening was actually real in the modified transverse scan, the fetus was examined in the dorsal midsagittal plane perpendicular to the nuchal area using axial resolution. A nuchal bulge was considered abnormal while a flat or slightly concave nuchal area was considered normal. Of interest, we described a new sonographic observation in the modified transverse view when there was true nuchal thickening (Ginsberg *et al*., 1990). There is a thickened curvilinear extension of the subcutaneous tissue and the overlying skin line as it approaches and covers a portion of the lateral skull due to edema of the subcutaneous layer ('lateral edema' sign). This finding has been documented without being recognized (Benacerraf and Frigoletto, 1987; Nyberg and Souter, 2001).

We suggest that a detailed dorsal midsagittal scan be obtained perpendicular to the nuchal area (using axial resolution) before rendering a final diagnosis in cases where the transverse view is ≥ 5 mm. The fetal head and neck should be in a neutral or near-neutral position between 14 and 20 weeks' gestation. A dorsal midsagittal view of a normal fetus shows a slightly concave-to-flat nuchal skin line (Figure 1). A dorsal midsagittal view of an abnormal fetus shows thickening of the subcutaneous tissue due mainly to edema with its associated characteristic smooth outward bulging (convexity) of the nuchal skin line (Figure 2). The abnormal fetus in this view usually demonstrates two adjacent sonographic zones in the region of nuchal thickening. The echogenic skin line overlies a hypoechoic stripe that represents edematous subcutaneous tissue. The muscles are ventral and an echogenic linear specular reflector overlying the muscles is probably due to fascia. With the head in neutral position, the skin bulges in the nuchal region even if the two zones cannot be seen. This is called the 'nuchal bulge' sign. The 'nuchal bulge' sign, as well as the 'lateral edema' sign, are beautifully displayed in the textbook *Ultrasound of Fetal Syndromes* by Benacerraf (1998) on pages 86 and 329.

There are at least four major advantages of the dorsal midsagittal view over the transverse view: first, the ultrasonographer can directly visualize the bulging of the nuchal skin line (Figure 2); second, the ultrasonographer can determine the position of the fetal head and neck at the exact time the scan is obtained (Figure 2); third, the ultrasonographer can obtain a detailed view of the fetal neck anatomy demonstrating the skin line delineating the dorsal aspect of the subcutaneous area, the subcutaneous tissue, a specular reflector attached to the lower occiput probably due to fascia delineating the ventral aspect of the subcutaneous area and the adjacent muscles (Figures 1 and 2); and four, the use of axial rather than lateral resolution allows better detail and more accurate measurements as well as demonstration of specular reflectors.

There are advantages of the transverse view because

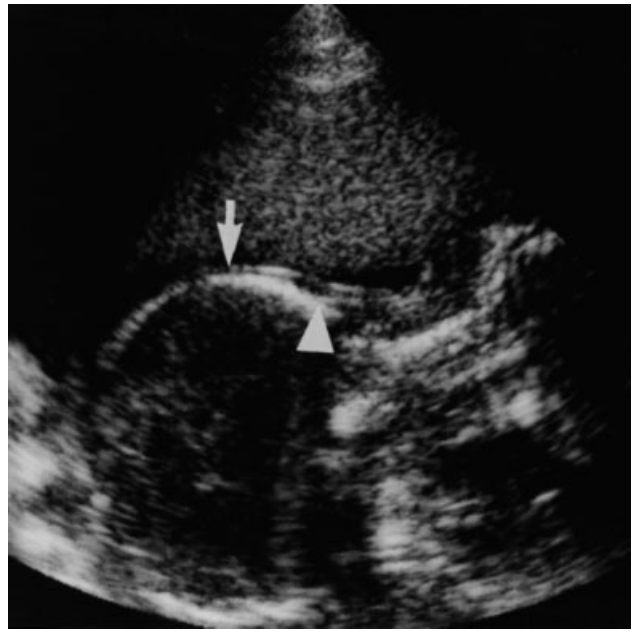

Figure 1—Dorsal midsaggital view of a normal fetus demonstrating the skin line (arrow) overlying the hypoechoic subcutaneous tissue and a specular reflector, probably representing a fascial plane (arrowhead) overlying the neck muscles. The arrow marks the upper occipital region while the arrowhead marks the lower occipital region where the fascia attaches. Notice the slight concavity of the nuchal skin line

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Figure 2—Dorsal midsaggital view of a Down syndrome fetus with the head and neck in neutral position showing thickening of the subcutaneous tissue and bulging of the nuchal skin line (nuchal bulge sign). The arrow and arrowhead indicate the same regions as in Figure 1

it is usually easier to obtain and ultrasonographers are more familiar with this view. The dorsal midsagittal view can be time-consuming if the fetal lie and position are unfavorable. The dorsal midsagittal view may also require unusual degrees of transducer pressure and angulation to obtain a detailed scan as well as optimal maternal bladder distention. Therefore, the transverse and dorsal midsagittal views should be considered complementary to one another.

The use of the modified transverse view in combination with the dorsal midsagittal view was clearly responsible for our false-positive rate of 0% in screening second trimester pregnancies for Down syndrome (Ginsberg *et al*., 1990). Since that time, over 8000 pregnancies between 14 and 20 weeks' gestation have been evaluated with this approach and only one false-positive occurred (rate = 0.0125%). Therefore, it is proposed that the dorsal midsagittal view become part of the standard and required methodology when screening for Down syndrome beyond the first trimester. This approach will reduce false-positive diagnoses, increase the positive predictive value, and, most importantly, reduce the number of unnecessary amniocenteses and subsequent loss of normal fetuses due to the inherent risks of amniocentesis.

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