Nomograms of Fetal Nuchal Fold Thickness for Early Detection of Down Syndrome

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Abstract. A prospective study was undertaken in North India to find the normative data on the nuchal fold thickness of randomly selected 150 fetuses between 16-22nd week of gestation. The data revealed that the width of nuchal fold thickness ranged between 2-5 mm. The mean for 16-18, >18-20, and >20-22 week's gestation was 2.6±0.2, 3.8±0.9, and 4.0±0.8 mm respectively. In one fetus nuchal fold thickness was 6mm at 18 weeks (>2SD above the normal value) and thus Down Syndrome was suspected. This was later confirmed by amniocentesis and karyotyping. (Indian J Pediatr 1993; 60 : 655-657)

Key words: Nuchal fold; Ultrasound; Down Syndrome; Prenatal diagnosis.

The incidence of Down Syndrome is approximately 1 in 660 live birth's,¹ and it is the commonest chromosomal malformation leading to mental retardation. Until recently the diagnosis of Down Syndrome was based on cytogenetic study of fetal cells or fetal tissue. Now sonography provides an alternative, non invasive technique for detection of Down Syndrome. Symmetrical, flat, excess, posterior nuchal thickness on ultrasonography during 2nd trimester was the first sonographic sign reported.² This finding can be taken as a screening procedure in all high risk mothers for Down Syndrome during 2nd trimester, and useful for identifying younger women who are also at significantly increased risk for having neonates with Down Syndrome, and to whom cytogenetic studies should therefore be offered.

Material and Methods
A total number of 150 randomly selected pregnant women attending the antenatal clinic at AIIMS were included in the study. Serial ultrasonography was done from 16th to 22nd week of gestation at fortnightly interval using a real time gray scale, ultrasound machine (Aloka echo camera SSD-256, model 3.5 MHZ). The fetal head was scanned in the transverse plane which included the thalamus, their ventricle and occipital tissue above the level of cerebellum. The distance from the external surface of the occipital bone to the external surface of the overlying skin was measured by electronic callipers (Figure 1). Caution was taken to exclude false positive reading by avoiding angled planes of the cerebellum, and not to measure the thickness from a plane taken below the level of occipital bone.

Results
The mean value of the nuchal fold thick-
ness in 150 fetuses between 16th to 22nd week of gestation is displayed in Table 1.

In one case the nuchal fold thickness was 6mm at 18th week of gestation (Figure 2), which was 2 SD above the normal for that period of gestation. Thus fetal trisomy was suspected and it was confirmed on karyotyping from amniocytes.

**DISCUSSION**

Only 25-30% of fetuses with Down Syndrome are born to women 35 years of age or older. But the majority of fetuses with Down Syndrome are born to younger women, most of whom are not screened for trisomy 21 during antenatal evaluation. Thus the unexpected arrival of a neonate with Down Syndrome leads to tremendous psychological stress to the obstetrician, couple and burden to the society. Therefore a simple, rapid, non invasive, screening test for Down Syndrome will be very helpful. Recent studies have indicated that a nuchal fold thickness >6 mm in the second trimester of pregnancy has been associated with Down Syndrome. Thus we studied fetuses (150) between 16-22 weeks of gestation to find out the norms of nuchal skin thickness in Indian fetuses. The range of nuchal fold thickness was 2-5 mm in fetuses between 16-22 weeks gestation. This is in accordance with Benacerraf’s study. In one fetus, the nuchal fold thickness was 6 mm at 18 weeks of gestation (2SD above the normal value). The fetus did not have any sign of hydrops fetalis. Thus Down syndrome was suspected, which was later confirmed by karyotyping from amniocytes. Benacerraf et al stated that

<table>
<thead>
<tr>
<th>Nuchal fold thickness (mm)</th>
<th>16-18 wk</th>
<th>&gt;18-20 wk</th>
<th>&gt;20-22 wk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mean (mm)</td>
<td>2.6</td>
<td>3.8</td>
<td>4.0</td>
</tr>
<tr>
<td>S.D.</td>
<td>0.2</td>
<td>0.9</td>
<td>0.8</td>
</tr>
</tbody>
</table>

**Fig 1.** Nuchal fold thickness - 3mm at 18 weeks (normal).

**Fig 2.** Nuchal fold thickness - 6mm at 18 weeks (Down syndrome).