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ORIGINAL ARTICLES

Ultrasonographic Femur-Tibial Length Ratio: A Marker of Down Syndrome from the Late Second Trimester

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ABSTRACT

An observational prospective study reported that newborn babies with Down syndrome (DS) had short upper limbs that reach up to their pelvis. The shortening was most marked in the forearm (the middle segment of the upper limb) and this relative shortening resulted in an alteration of the proximal to middle segment length ratio. This study assumes that there is a similar alteration in the ratio of the lower limb. We propose to study the proximal to middle segment ratio in the lower limb in normal fetuses at different gestational ages. Against these norms we propose to study the ratio in fetuses with DS to see at what stage in intrauterine life the altered ratio becomes evident. We also propose to take postnatal measurements of upper and middle segments of both upper and lower limbs of babies born with DS and
compare them with normal babies. Fetal femoral and tibial lengths were measured by routine antenatal ultrasound scans at a General hospital with 6000 deliveries a year. All babies delivered were examined for phenotypical evidence of DS. The in utero measurements recorded of babies born with DS were compared with the measurements in normal babies. Postnatal measurements of the arm and forearm, and the thigh and leg of babies with DS were taken soon after birth. These were compared with a control group of 20 consecutive normal babies born over 2 days. There were 3690 readings of 3075 normal fetuses and 8 measurements of 7 Down fetuses. The leg, the upper arm, and arm of newborns with DS were significantly shorter than controls ($p < 0.01$). The upper limb reached up to the pelvis in infants with DS and not up to mid thigh as in normal babies. The forearm was shorter than the arm in infants with DS. This is a reversal of the ratio seen in controls. The ratio of femoral to tibial length remains near constant at 1.1 after 13 weeks' gestation in normal fetuses. It rises from 1.2 to 1.4 from 22 weeks' to 38 weeks' gestation in fetuses with DS. The mean standard deviation score of fetuses with DS was 4.53 compared with norms ($SD$ $1.7$, $p < 0.01$). Conclusions of this study are: (1) short upper limbs (reaching only up to the pelvis) is a clinical feature of DS at birth; and (2) after 20 weeks' gestation, the ratio of femoral-tibial length can be a marker of DS in utero.

**KEYWORD**

Trisomy 21 - antenatal diagnosis - fetal limb length