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UPDATES IN CLINICAL MEDICINE

Update in Obstetrics and Gynecology

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It is now clear that most of the major fetal abnormalities can be diagnosed prenatally by ultrasound, and most important, that most of these abnormalities can be detected in the first trimester of pregnancy, using some easily recognizable markers. Good examples of such markers are the scalloping of the frontal bones (the 'lemon' sign) and caudal displacement of the cerebellum (the 'banana' sign), observed in the second trimester in most fetuses with open spina bifida.

A major challenge in first-trimester ultrasonography has been the diagnosis of open spina bifida. This challenge, however, may now have been solved by the realization that open spina bifida can be suspected by an easily detectable marker within the brain in the same mid-sagittal plane of the fetal face as for measurement of the NT and assessment of the nasal bone. In normal fetuses the fourth cerebral ventricle presents as an intracranial translucency (IT) parallel to the NT, while in fetuses with open spina bifida there may be absence of the IT.

Measurement of IT is similar to that of NT. The two lines that define the IT are the posterior border of the brain stem anteriorly and the choroid plexus of the fourth ventricle posteriorly. Its measurement is similar to that of NT. The exact mid-sagittal plane of the fetal face should be obtained and the image should be magnified so that only the fetal head and upper thorax are included. The exact mid-sagittal plane of the fetal face is defined by the echogenic tip of the nose and rectangular shape of the palate anteriorly, the translucent thalamus in the center and the nuchal membrane posteriorly. At 11–13 weeks the brain stem appears hypoechogenic (dark gray) whereas the IT is anechoic (black).

The optimal gestational age for measurement of fetal NT is 11 + 0 to 13+ 6 weeks. At 11–13 weeks it is possible to diagnose severe brain abnormalities, including holoprosencephaly, ventriculomegaly, acrania-exencephaly and encephalocele.

Within the gestational age range of 11–13 weeks the antero-posterior diameter of the IT increases with fetal crown–rump length (CRL) from a median of 1.5 mm at a CRL of 45 mm to 2.5 mm at a CRL of 85. The fourth ventricle can be identified easily after 13 weeks and the diameter increases with gestational age.

The fourth cerebral ventricle can be visualized both trans-abdominally and trans-vaginally; measurement of IT is best carried out in the midsagittal plane of the fetal face, which is easier to obtain trans-abdominally.

Three-dimensional ultrasound is useful in assessing IT particularly when it is difficult to obtain the mid-sagittal plane directly by two-dimensional ultrasound. In such cases a transverse view of the fetal head at the level of the

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fourth ventricle is obtained. This is best achieved by trans-vaginal sonography because the resolution is higher.

In the 1980s, the main method of screening for open spina bifida was by maternal serum α-fetoprotein at around 16 weeks and the method of diagnosis was amniocentesis and measurement of amniotic fluid α-fetoprotein and acetyl cholinesterase.

Although it was possible to diagnose the condition by ultrasonographic examination of the spine, the sensitivity of this test was low. Generally, most of the cases were diagnosed with second-trimester ultrasonography.

In the 1990s the emphasis shifted to the first trimester when it was realized that the great majority of trisomic fetuses have increased NT that can be detected easily in a mid-sagittal plane of the fetal face at 11–13 weeks.

It is now clear that in this same mid-sagittal plane the fourth cerebral ventricle is easily visible as an IT and that at least in some cases of open spina bifida, caudal displacement of the brain is evident from the first trimester. If this is not visible the sonographers will be alerted to the possibility of an underlying open spina bifida and will undertake detailed examination of the fetal spine.

Prospective large studies will determine the proportion of affected fetuses presenting with absent IT and the extent to which the 11–13-week scan can provide an effective method for early diagnosis of open spina bifida.

References

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For further information on this topic, you may address The *Ultrasound Obstet Gynecol* 2010; 35: 133–138, Editorial, From nuchal translucency to intracranial translucency: towards the early detection of spina bifida; authors: **R. CHAOUI** and **K. H. NICOLAIDES**.