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# Assessment of intracranial translucency (IT) in the detection of spina bifida at the 11–13-week scan

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**KEYWORDS:** first trimester; nuchal translucency; prenatal diagnosis; spina bifida; ultrasound

## ABSTRACT

***Objective*** *Prenatal diagnosis of open spina bifida is carried out by ultrasound examination in the second trimester of pregnancy. The diagnosis is suspected by the presence of a 'lemon-shaped' head and a 'banana-shaped' cerebellum, thought to be consequences of caudal displacement of the hindbrain. The aim of the study was to determine whether in fetuses with spina bifida this displacement of the brain is evident from the first trimester of pregnancy.*

***Methods*** *In women undergoing routine ultrasound examination at 11–13 weeks' gestation as part of screening for chromosomal abnormalities, a mid-sagittal view of the fetal face was obtained to measure nuchal translucency thickness and assess the nasal bone. In this view the fourth ventricle, which presents as an intracranial translucency (IT) between the brain stem and choroid plexus, is easily visible. We measured the anteroposterior diameter of the fourth ventricle in 200 normal fetuses and in four fetuses with spina bifida.*

***Results*** *In the normal fetuses the fourth ventricle was always visible and the median anteroposterior diameter increased from 1.5 mm at a crown–rump length (CRL) of 45 mm to 2.5 mm at a CRL of 84 mm. In the four fetuses with spina bifida the ventricle was compressed by the caudally displaced hindbrain and no IT could be seen.*

***Conclusion*** *The mid-sagittal view of the face as routinely used in screening for chromosomal defects can also be used for early detection of open spina bifida. Copyright © 2009 ISUOG. Published by John Wiley & Sons, Ltd.*

## INTRODUCTION

In the 1980s the main method of screening for open spina bifida was by maternal serum α-fetoprotein at around 16 weeks of gestation, 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[1,2]. However, the observation that spina bifida was associated with scalloping of the frontal bones (the 'lemon sign') and caudal displacement of the cerebellum (the 'banana sign'), has led to the replacement of biochemical assessment with ultrasonography, both for screening and for diagnosis of this abnormality[3].

In the last 10 years there has been widespread uptake of routine ultrasound examination in the first trimester of pregnancy, and in the UK the National Screening Committee has recommended that this scan should be offered to all pregnant women. The 11–13-week scan is used for measurement of the fetal crown–rump length (CRL) to determine gestational age, for diagnosis of major abnormalities such as anencephaly and to screen for trisomy 21 and other aneuploidies. The latter relies on accurate measurement of the fetal nuchal translucency (NT) thickness and assessment of the nasal bone, which necessitates examination of the mid-sagittal view of the fetal face[4,5].

Extensive studies have reported that in addition to aneuploidies the 11–13-week scan can identify the majority of all major fetal abnormalities[6]. However, in the case of spina bifida the diagnosis is usually missed at this scan. A screening study at 11–13 weeks in 61 972 pregnancies undergoing measurement of fetal NT reported that none of the 29 fetuses with spina bifida was detected[7].

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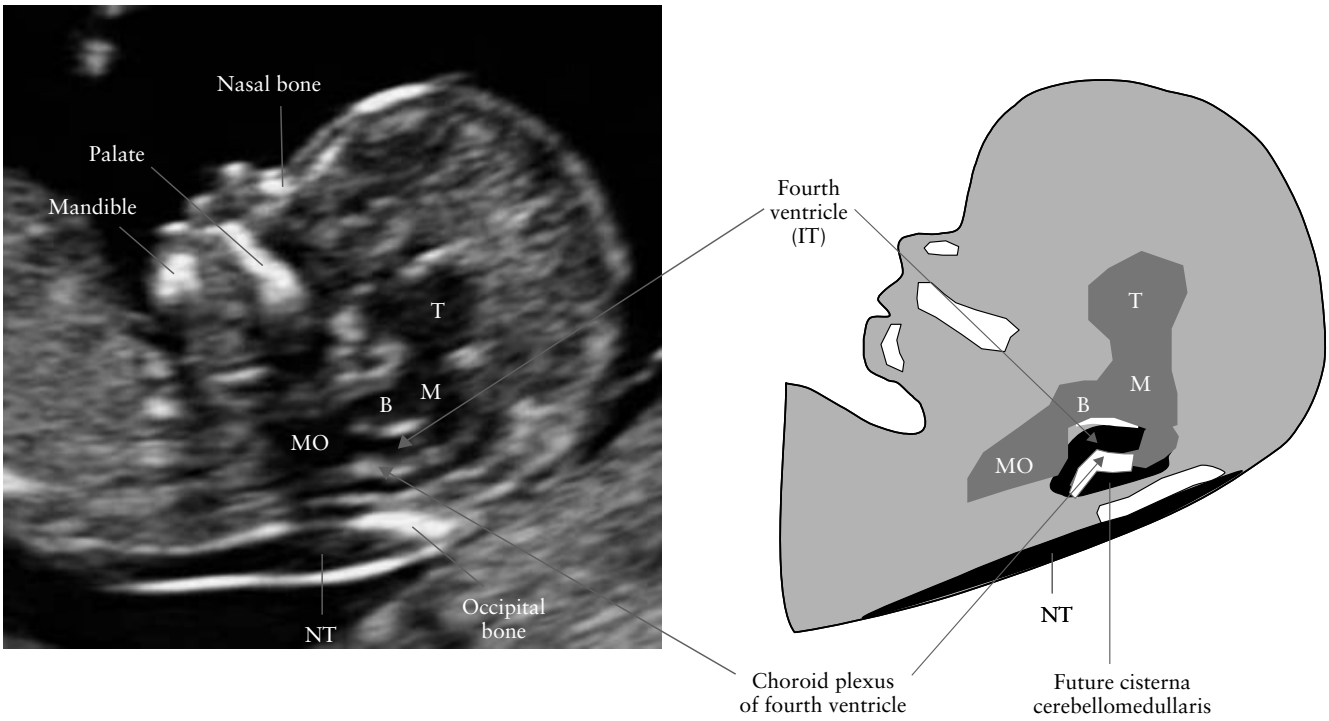


**Figure 1** Ultrasound image in the mid-sagittal plane of the fetal face showing the nasal bone, palate, mandible, nuchal translucency (NT), thalamus (T), midbrain (M), brain stem (B) and medulla oblongata (MO). The fourth ventricle presents as an intracranial translucency (IT) between the brain stem and the choroid plexus.

In the same mid-sagittal view of the fetal face as used for measurement of NT and assessment of the nasal bone, the brain stem and fourth cerebral ventricle are easily visible (Figure 1). The fourth ventricle presents as an intracranial translucency (IT) parallel to the NT and is delineated by two echogenic borders; the dorsal part of the brain stem anteriorly and the choroid plexus of the fourth ventricle posteriorly. Between the fourth ventricle and the occiput there is another thinner translucency generated by the developing cisterna cerebellomedullaris.

The aim of the study was to determine whether in fetuses with spina bifida the associated caudal displacement of the brain resulting in compression of the fourth ventricle is evident from the first trimester of pregnancy.

## PATIENTS AND METHODS

In our centers we perform first-trimester screening for chromosomal defects by a combination of measurements of levels of maternal serum free β-human chorionic gonadotropin and pregnancy-associated plasma protein-A with ultrasound findings at 11–13 weeks' gestation. As part of the scan we obtain the mid-sagittal view of the fetal face, as recommended by The Fetal Medicine Foundation, for measurement of fetal NT and assessment of the nasal bone. In all cases we store the image of the mid-sagittal view of the face electronically.

We searched our database to identify cases of spina bifida diagnosed at the first- or second-trimester scan and 200 consecutively examined normal fetuses with stored images of the mid-sagittal view of the fetal face at 11–13 weeks. The images were examined by two operators who were not aware of the diagnosis and were asked to identify the fourth ventricle. In addition, one of the operators measured the anteroposterior diameter using the electronic calipers of the machine. Regression analysis was used to determine the significance of the association between the anteroposterior diameter of the fourth ventricle and CRL.

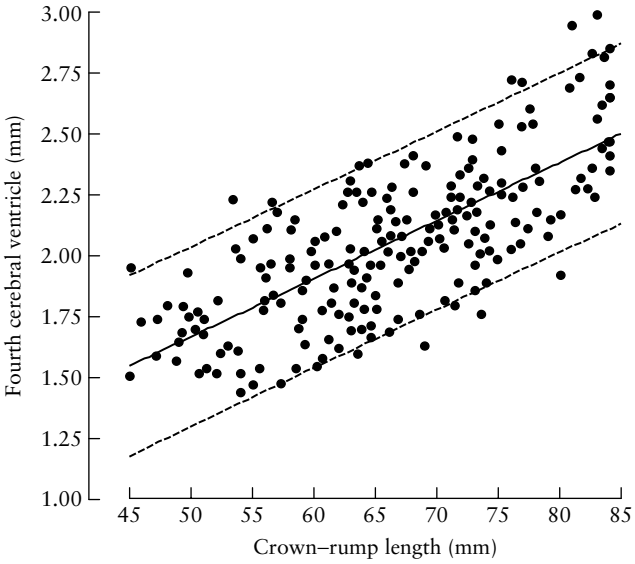


**Figure 2** Reference range (mean, 5th and 95th centiles) of fourth ventricle anteroposterior diameter according to crown–rump length.

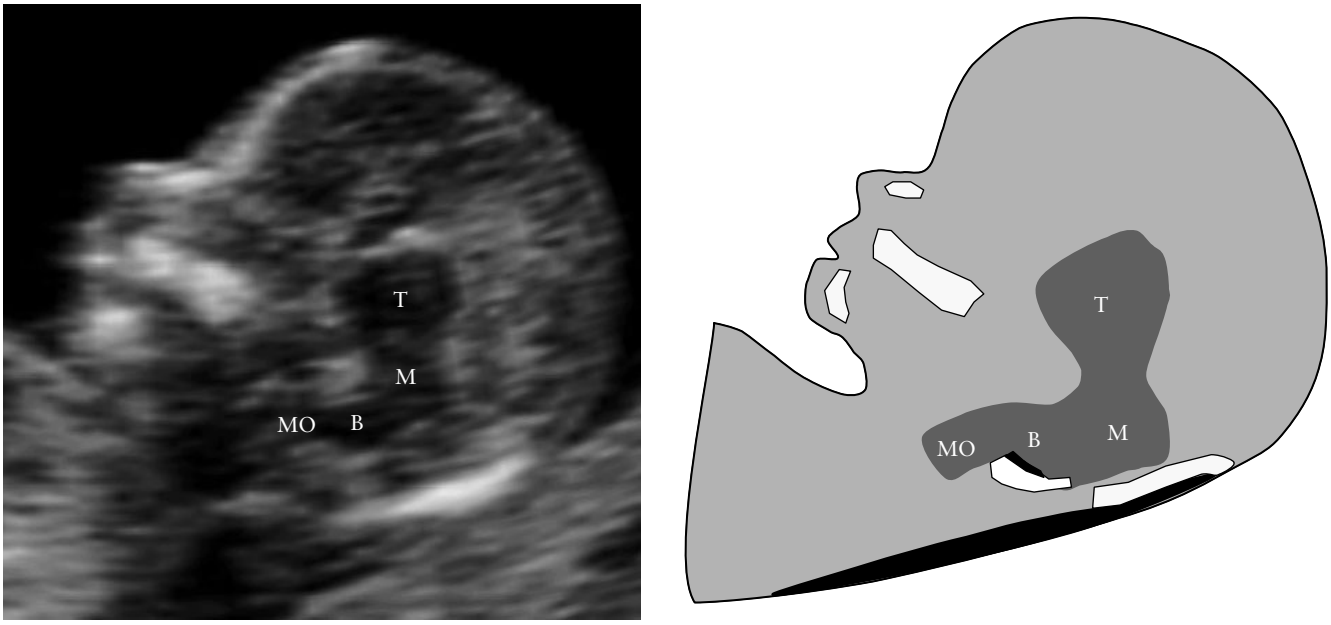


**Figure 3** Ultrasound image in the mid-sagittal plane of the fetal face in a case of open spina bifida demonstrating compression of the fourth ventricle with no visible translucency. B, brain stem; M, midbrain; MO, medulla oblongata; T, thalamus.

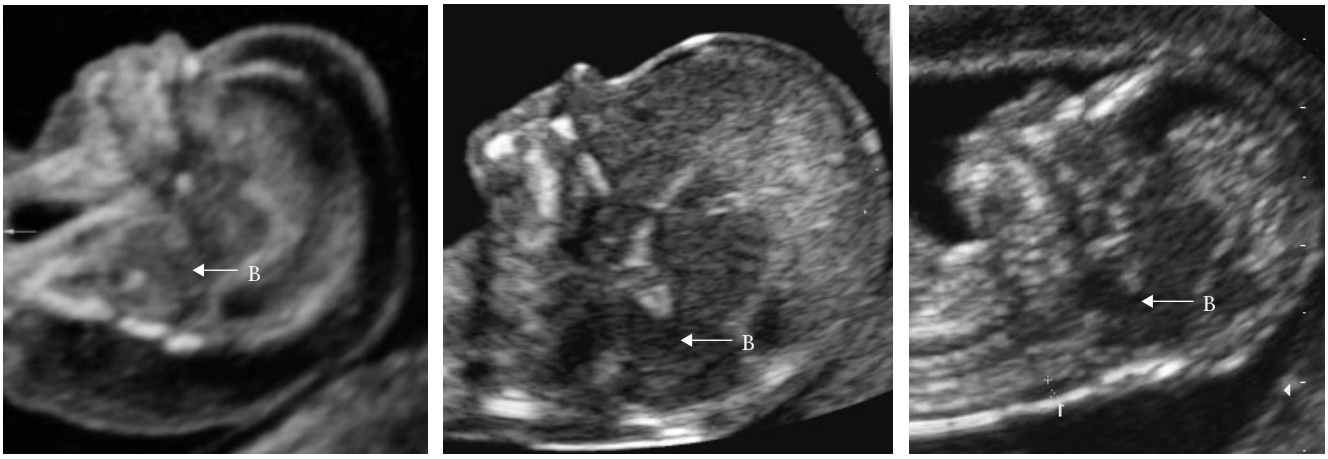


**Figure 4** Ultrasound images in the mid-sagittal plane of the fetal faces of the three additional cases of spina bifida. The arrows point to the brain stem with absence of the fourth ventricle (compare with the normal case in Figure 1).

## RESULTS

In the normal fetuses the median CRL was 65 (range, 45–84) mm and the median gestation was 12 (range, 11–13) weeks. Both operators easily identified the fourth ventricle in all cases. The anteroposterior diameter of the fourth ventricle increased linearly with gestation from a median of 1.5 mm at a CRL of 45 mm to 2.5 mm at 84 mm ($r = 0.736$, $P < 0.0001$; Figure 2).

In the four cases of spina bifida the CRL at 11–13 weeks was 53 mm, 55 mm, 60 mm and 76 mm, respectively. There was agreement by both operators that, in the mid-sagittal view of the fetal face, the fourth cerebral ventricle was not visible in any of the cases (Figures 3 and 4). The diagnosis of open sacral spina bifida was made in the second trimester and in all cases the 'lemon' and 'banana' signs were present.

## DISCUSSION

The findings of this study demonstrate that at 11–13 weeks' gestation the fourth cerebral ventricle is easily recognizable as an intracranial translucency in the standard mid-sagittal view of the face used routinely in screening for chromosomal abnormalities. The data also suggest that at least in some cases of open spina bifida the fourth ventricle is not visible.

In almost all cases of open spina bifida there is an associated Arnold–Chiari malformation, which is thought to be a consequence of the leakage of cerebrospinal fluid into the amniotic cavity and hypotension in the subarachnoid spaces leading to caudal displacement of the brain and obstructive hydrocephalus[8,9]. In the second trimester of pregnancy the manifestations of the Arnold–Chiari malformation are the 'lemon' and 'banana' signs[3]. Our

findings suggest that in open spina bifida caudal displacement of the brain is evident from the first trimester, resulting in compression of the fourth ventricle and loss of the normal intracranial translucency.

Examination of the mid-sagittal view of the fetal face is performed routinely for assessment of fetal NT and the nasal bone in screening for aneuploidies. If in this same view the fourth ventricle is not visible the sonographer should be alerted to the possibility of an underlying open spina bifida and undertake detailed examination of the fetal spine. Prospective large studies are necessary to determine the performance of intracranial translucency in screening for open spina bifida.

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