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REVIEW ARTICLE

Ultrasonographic Evaluation of Neural Tube Defects at 11–14 Weeks

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Abstract Fetal neural tube defects are the second most common serious fetal birth defects. In fetuses with open spina bifida there is leakage of cerebrospinal fluid into the amniotic cavity which causes hypertension in the subarachnoid space leading to caudal displacement of the brain. The cranial signs (lemon and banana) are detectable during the second trimester scan. In the first trimester, there are alterations in the appearance of the posterior brain at 11–13 weeks. In fetuses with open spina bifida, the posterior shift of the brain is evident from the first trimester, resulting in compression of the fourth ventricle and alteration of the normal intracranial translucency. In the midsagittal view of the face, if the fourth ventricle is not visible, it should arouse the suspicion of the possibility of an underlying open spina bifida and hence, a detailed examination of the fetal spine should be performed. Also, because of the posterior shift of the brain, the brainstem (BS) appears relatively enlarged and shifted backwards. The BS to brainstem to occipital bone distance (BSOB) [BS/BSOB] ratio is increased and it is usually more than one. At 11–13-week scan, the midsagittal view of the face is the standard view obtained in every fetus and it is feasible to visualize the posterior region of brain in this view. Suspicious cases should undergo a detailed assessment of the spine, especially on transvaginal scan followed by targeted examination of the spine at 16–18 weeks.

Keywords Neural tube defects · Posterior fossa · Intracranial translucency · Brainstem to occipital bone distance · Transvaginal scan

Introduction

Fetal neural tube defects (NTDs) are the second most common serious fetal birth defect, surpassed only by congenital heart defects. Screening methods used to identify NTDs are now a component of routine obstetrical care, and include both second trimester maternal serum alpha-fetoprotein (MSAFP) levels and fetal ultrasonographic evaluations. Once an NTD is identified, various management options are available for families, including consideration of pregnancy termination, in utero fetal surgery, as well as referral to a tertiary care center for management and delivery. Therefore, it is useful for the practicing obstetricians to be aware of these complex diagnostic and management options for optimal care of the patient and fetus [1].

Posterior Fossa at 11–14 Weeks

In fetuses with open spina bifida, there is leakage of cerebrospinal fluid (CSF) into the amniotic cavity, which causes hypertension in the subarachnoid space leading to caudal displacement of the brain, and it is this caudal displacement of brain that is seen as the lemon sign because of scalloping of the frontal bones and also as the banana sign because of the caudal displacement of the cerebellum.

These cranial signs are easily detectable during the second trimester scan. In the first trimester, however, these

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signs cannot be relied upon for the diagnosis of open spina bifida. In recent years, there have been preliminary reports that suggest alterations in the appearance of the posterior brain at 11–13 weeks in fetuses with open spina bifida [2–4].

Midsagittal View

The midsagittal plane of the face is routinely evaluated at 12 weeks to screen the fetus for aneuploidy by measuring the nuchal translucency as well as assessing the nasal bone. The posterior brain can be evaluated in the same plane. At 11–13 weeks, the fourth ventricle presents as an intracranial translucency (IT) that appears as a fluid-filled translucent region with two echogenic horizontal borders:

(a) Posterior border of the brainstem (BS) anteriorly.
(b) Choroid plexus of the fourth ventricle posteriorly (Fig. 1).

The fluid of the future cisterna magna is readily identified between the choroid plexus and the occipital bone. There is a progressive increase in IT from 1.5 to 2.5 mm at a crown rump length (CRL) of 45–85 mm. But unlike nuchal translucency (NT), in which we are so stringent about always getting the midsagittal plane, there is very minimal variation seen in the measurement of IT even when you go a little off-axis.

Recent studies have shown that in fetuses with open spina bifida, there is caudal displacement of the brain, which also causes the posterior shift of the brain that is evident from the first trimester, resulting in compression of the fourth ventricle and alteration of the normal IT. The midsagittal view of the face is routinely evaluated in the first trimester. So in the same view, if the fourth ventricle is not visible, it should arouse the suspicion of the possibility of an underlying open spina bifida, and hence, a detailed examination of the fetal spine should be performed [2]. If the examination of the spine at 12 weeks appears to be normal, a detailed evaluation of the spine with the help of transvaginal scan followed by an ultrasound study at 16 and 20 weeks with particular attention to the spine should be performed.

A study was done by a group in Toronto [5] in which, three examiners, blinded to fetal outcome, analyzed images from a database of 199 fetuses, including eight with open spina bifida. In 150 images, a clear IT was seen and all of these fetuses were normal. In the remaining 49, the IT was not clearly seen, either for technical reasons (n = 43) or because the examiner considered it a possible case of spina bifida (n = 6). Interestingly, there were four fetuses with spina bifida in each of these two groups. This study shows that a normal IT and posterior brain region have a high specificity for exclusion of spina bifida, which is a prerequisite for a good screening test. So if there is a normal IT, there is a high chance that there is no open spina bifida at 12-week scan. So in most cases of open spina bifida, the fourth ventricle is not visible. But sometimes some amount of fluid is seen, which might be still present posterior to the BS without the typical landmarks of the IT.

So, in the midsagittal view of the brain, three echogenic lines are seen. The first one is formed by the posterior border of the BS, the second line is formed by the choroid plexus of the fourth ventricle, and the third line formed by the occipital bone (Fig. 2).

In the midsagittal plane itself, the lower part of the fetal brain between the sphenoid bone anteriorly and the occipital bone posteriorly can be divided into the BS and the area that is posterior to it, which is a combination of the

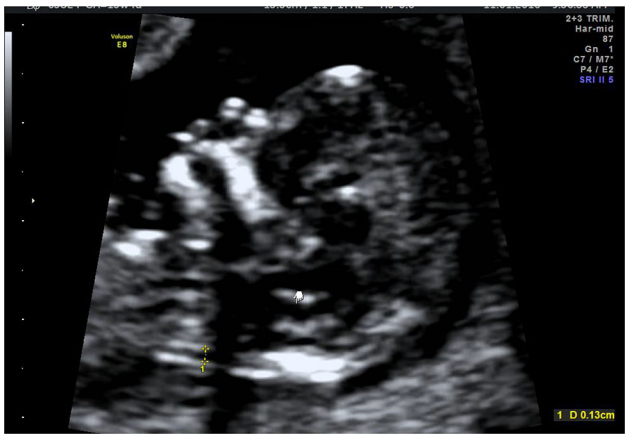

Fig. 1 Ultrasound image in the midsagittal plane of the fetal face showing the nasal bone, palate, mandible, and nuchal translucency. The fourth ventricle presents as an intracranial translucency (*pointer*) between the brainstem and the choroid plexus

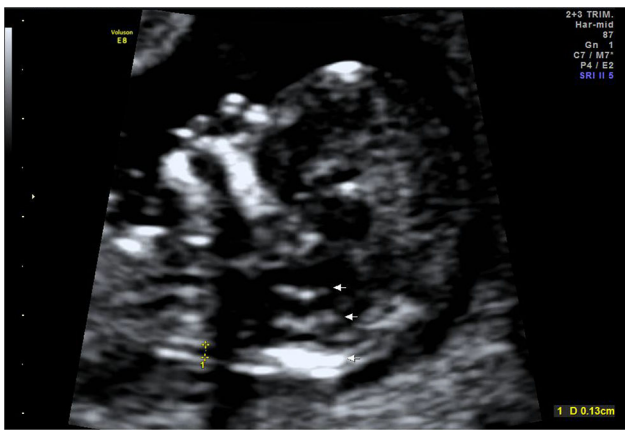

Fig. 2 Ultrasound image showing three echogenic *parallel lines* in the posterior brain (*arrows*)

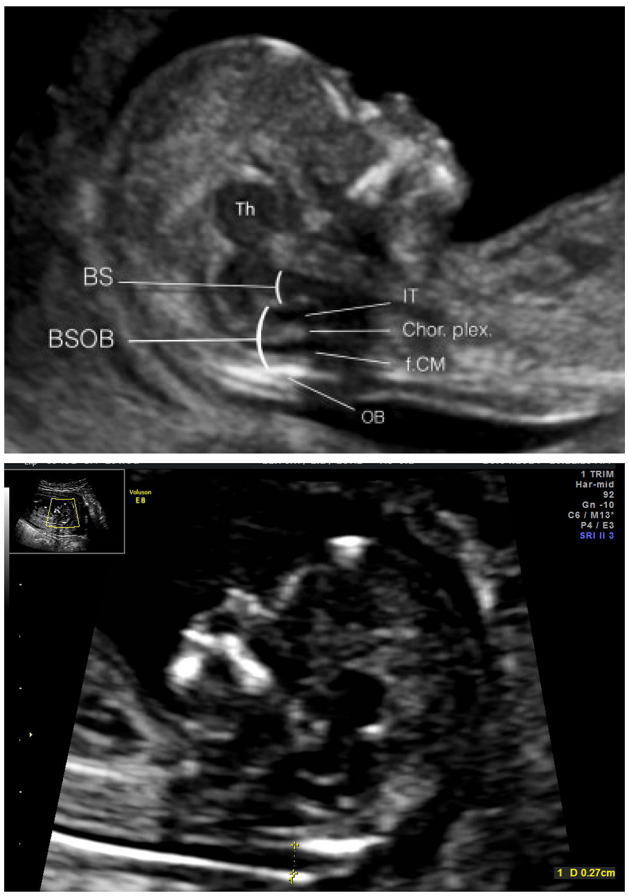

Fig. 3 Ultrasound image showing landmarks for brainstem and brainstem to occipital bone distance

fourth ventricle and cisterna magna in the back also referred to as brainstem to occipital bone distance (BSOB) (Fig. 3). Normally, the BS is always smaller than the area that is posterior to it and when there is an open spina bifida, because of the posterior shift of the brain, this BSOB will be lesser and the BS would be relatively larger, whereas in the fetuses with open spina bifida, the BS appears to be much more widened because of the posterior shift and it is relatively much larger than the area that is posterior to it.

So normally, the ratio of the BS to the BSOB will range from 0.8 at 11 weeks to somewhere around 0.7 at 14 weeks. In fetuses with open spina bifida, because of the posterior shift of the brain, the BS appears relatively enlarged and shifted backwards, so the BS to BSOB (BS/BSOB) ratio is increased and it is usually more than one [4] (Fig. 4).

Axial View

The axial plane is routinely examined at 12 weeks to demonstrate the normal shape of the head, for measurement of the biparietal diameter, and to exclude the structural anomalies. But to look at the posterior fossa, we need to look at the axial plane in a slightly oblique fashion (axial oblique plane) something similar to imaging of the transcerebellar plane at 20-week scan.

The axial views are comparatively much more difficult to get and usually we can get them with much more ease with the help of a transvaginal scan rather than transabdominal scan. In this plane, the midbrain is visualized just caudal to the plane in which the biparietal diameter is measured. In the axial oblique plane, we see the two thalami with third ventricle in between, and this constitutes the diencephalon. Posterior to thalami are the two cerebral peduncles with the aqueduct of Sylvius (AOS) in between (Fig. 5). Under normal conditions, transition from the cerebral peduncles to the thalami follows an acute angle and there is some distance between the AOS and the occipital bone.

In fetuses with open spina bifida, due to the posterior shift of the midbrain, the cerebral peduncles distort and become juxtaposed to the occipital bone, with parallelism of the cerebral peduncles. The AOS at this gestation is seen

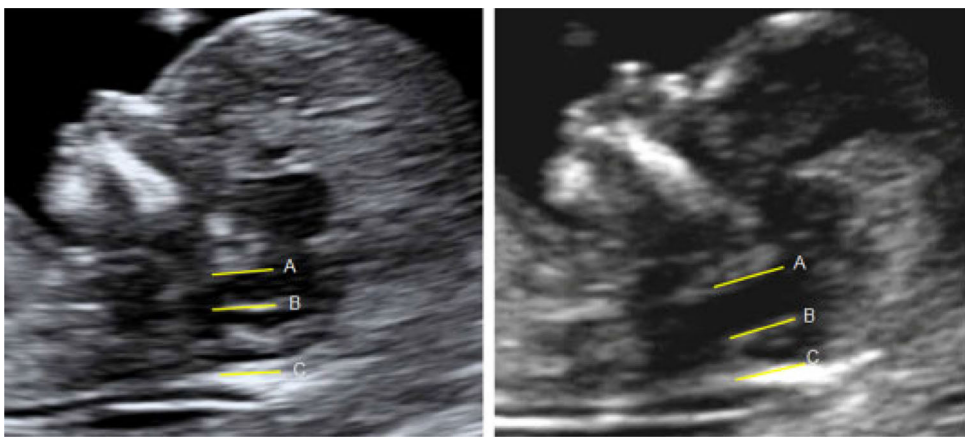

Fig. 4 Midsagittal view of the fetal brain showing the landmarks for measurement of brainstem diameter (between *Line A* and *Line B*) and brainstem to occipital bone distance (between *Line B* and *Line C*). *Line A* is drawn along the posterior border of the sphenoid bone, *Line B* along the anterior border of fourth ventricle, and *Line C* along the anterior border of the occipital bone (image courtesy—Lachmann et al.)

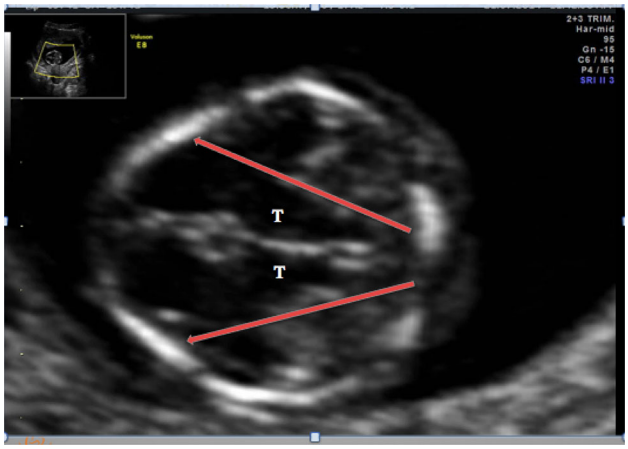

Fig. 5 Ultrasound image showing the two thalami (T) and the two cerebral peduncles posteriorly with the aqueduct of Sylvius in between

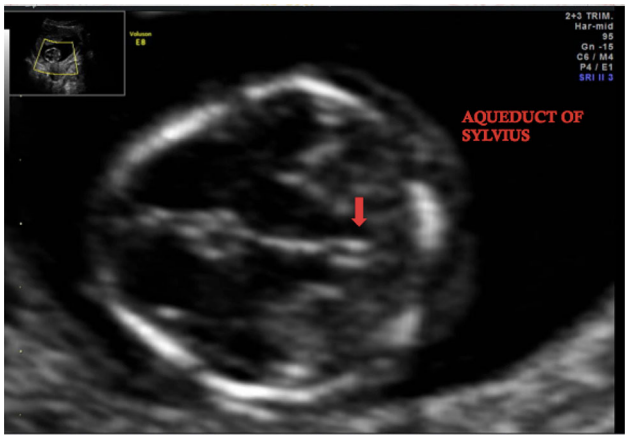

Fig. 6 Ultrasound image showing the aqueduct of Sylvius (*red arrow*)

as a prominent 'echogenic box' traversing the midbrain (Fig. 6).

It has also been observed that there is juxtaposition of the midbrain to the occipital bone, and quantification of this observation is done by measuring the distance between the AOS and the occipital bone, which is found to be reduced (Fig. 7). The AOS to occiput distance is measured from the posterior border of the AOS to the anterior border of the occiput in the axial plane (inner to inner) and it ranges from 1.7 mm (at CRL of 45 mm) to 3.7 mm (at CRL of 84 mm). This distance will be reduced in fetuses with open spina bifida because of juxtaposition of the midbrain to the occiput [6].

'Single-line' sign is another sign that has been described for evaluation of open spina bifida. A 3D volume has to be acquired in a midsagittal plane, preferentially through the anterior fontanelle. Axial sections are obtained along the z-axis just above and at the level of the upper part of the sphenoid bone. In normal fetuses, the cisterna magna is seen posterior to the fourth ventricle in the midsagittal section, and extends along the entire length. In the tilted axial view, there are two distinct transverse echogenic lines seen, an anterior line caused by the posterior border of the BS and a posterior line caused by the choroid plexus and the roof of the fourth ventricle. In contrast, in the fetuses with open spina bifida, the cisterna magna is obliterated in the cranial part and in the correct plane of the tilted axial view above the sphenoid bone, the cisterna magna is tiny or not visible. This results in the disappearance of posterior transverse echogenic line in the tilted axial view, resulting in the 'single-line' sign [7] (Fig. 8).

Another sign that has been described for detection of NTDs by 12-week scan is reduced frontomaxillary facial angle because basically, there is caudal displacement of the brain. This will lead to an impaired development of the frontal bones, which is responsible for the lemon sign at 12 weeks [8].

There are other studies, which have shown that in fetuses with open spina bifida, the biparietal diameter will be usually smaller (less than 10th centile) for the gestational age [9]. Loureiro et al. observed reduction of cerebral ventricular system at 12 weeks when there is open spina bifida. The reason for this is that the cerebrospinal fluid leaks out, reducing the amount of cerebrospinal fluid

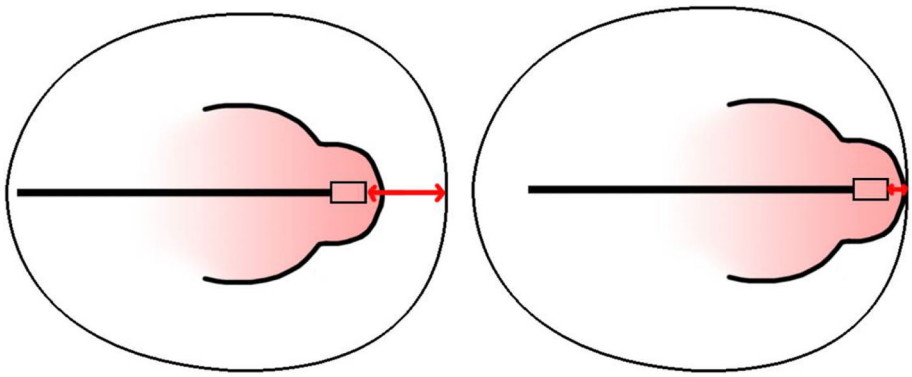

Fig. 7 Diagram showing the juxtaposition of the aqueduct of Sylvius with the occipital bone

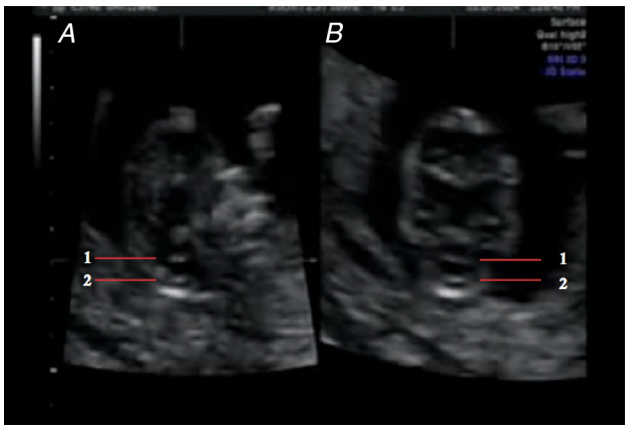

Fig. 8 3D ultrasound image showing *A* (sagittal) and *B* (axial oblique) plane. *Line 1* represents the posterior border of brainstem. *Line 2* represents the choroid plexus of fourth ventricle

in the intracranial area, which reduces the diameter of all ventricular cavities [10].

Conclusion

A screening test should be easy to perform and should require little additional effort over that routine examination. At 11–13-week scan, the midsagittal view of face is the standard view obtained in every fetus and it is feasible to visualize the posterior brain region. Suspicious cases should undergo a detailed assessment of spine, especially on transvaginal ultrasound scan followed by targeted examination of spine at 16–18 weeks. A more comprehensive examination should be performed by a specialist and is recommended in cases considered to be at high risk either because of a suspicious finding during routine screening or because of a history of previously affected pregnancies.

Compliance with Ethical Standards

Conflict of interest None.

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