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Opinion

Detecting open spina bifida at the 11–13-week scan by assessing intracranial translucency and the posterior brain region: mid-sagittal or axial plane?

Introduction

Prenatal diagnosis of open spina bifida in the second trimester can be achieved by identification of indirect cranial and cerebellar signs (lemon and banana signs), rather than by direct examination of the spine to locate the lesion[1]. Screening at 11–13 weeks' gestation is now performed not only for the measurement of nuchal translucency (NT), but also for detecting severe malformations and identifying pregnancies at high risk for adverse fetal and maternal outcome[2]. In the first trimester, open spina bifida has not been amenable to diagnosis because the banana and lemon signs cannot be relied upon at this gestational age[3,4]. However, we have recently reported that fetuses with open spina bifida have easily detectable abnormalities in the posterior fossa of the brain, which could be incorporated into the routine 11–13-week fetal sonographic assessment[5,6]. Our observation has been confirmed by several retrospective and prospective studies, many of which are reported in this issue of the Journal[7–15].

In our original report, examination of the posterior fossa was undertaken in the mid-sagittal plane that is used routinely for measurement of fetal NT and assessment of the nasal bone[5]. However, some studies have suggested that more reliable assessment of the posterior fossa and diagnosis of open spina bifida can be achieved using axial planes of the fetal head[9,16–18]. In this Opinion we discuss whether assessment of the posterior brain area should be undertaken in the mid-sagittal or axial plane.

Mid-sagittal plane: easiest and best approach

The rationale for using the mid-sagittal plane for examination of the posterior brain is that this view is obtained routinely in early screening for aneuploidies. The midbrain is one of the landmarks used to confirm the mid-sagittal plane and while attempting measurement of NT and assessment of the nasal bone the examiner can easily assess the structures of the posterior brain region.

At 11–13 weeks the fourth ventricle appears as a fluid-filled translucent region with two echogenic horizontal borders, representing the posterior border of the brainstem anteriorly and the echogenic thin choroid plexus of the fourth ventricle posteriorly (Figure 1a)[5–7]. The fluid of the future cisterna magna is readily identified between the choroid plexus and the occipital bone.

In our first report of four cases with spina bifida, we observed that in at least some cases of open spina bifida the fourth ventricle is not visible[5]. Further observations on a larger number of fetuses showed, however, that in some cases some fluid can still be present in the posterior brain, but without the typical landmarks of a normal intracranial translucency (IT) (Figure 1b)[7,11]. In an analysis of 30 cases with open spina bifida we observed, as a sign of fluid leakage and gradual development of Arnold–Chiari II malformation, a shift of the posterior brain towards the occipital bone. This was reflected by a thickening of the brainstem, a shortening of the distance between brainstem and occipital bone and an increase in the ratio of brainstem diameter to brainstem–occipital bone distance to more than 1 (normal value at 11–13 weeks is < 0.9; Figure 1b)[11].

Other groups have reported their experience of visualizing the IT under normal conditions and in cases with open spina bifida, confirming the feasibility of integrating this additional sign into the routine 11–13-week scan. Fong *et al.*[10], whose findings are published in this issue, conducted a study 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 recognized 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. Another study on quality control presented in this issue examines the ability of two experienced observers to successfully visualize the IT retrospectively on stored images from NT screening and on prospectively obtained images after specific training of the sonographers to visualize IT[15]. Whereas in the evaluation of the retrospective series of images the IT could be identified by both observers in only 52% of cases, this rate increased to 85% following specific training of the sonographers. The authors propose for future studies a score including eight criteria to assess the normality of the posterior brain and the IT, which is similar to what we describe in Figure 1a.

The incidence of open spina bifida is 1 in 2000 or less[4]. Successful screening for such an uncommon condition can only be achieved with a simple sign which can be visualized without excessive additional effort. In our

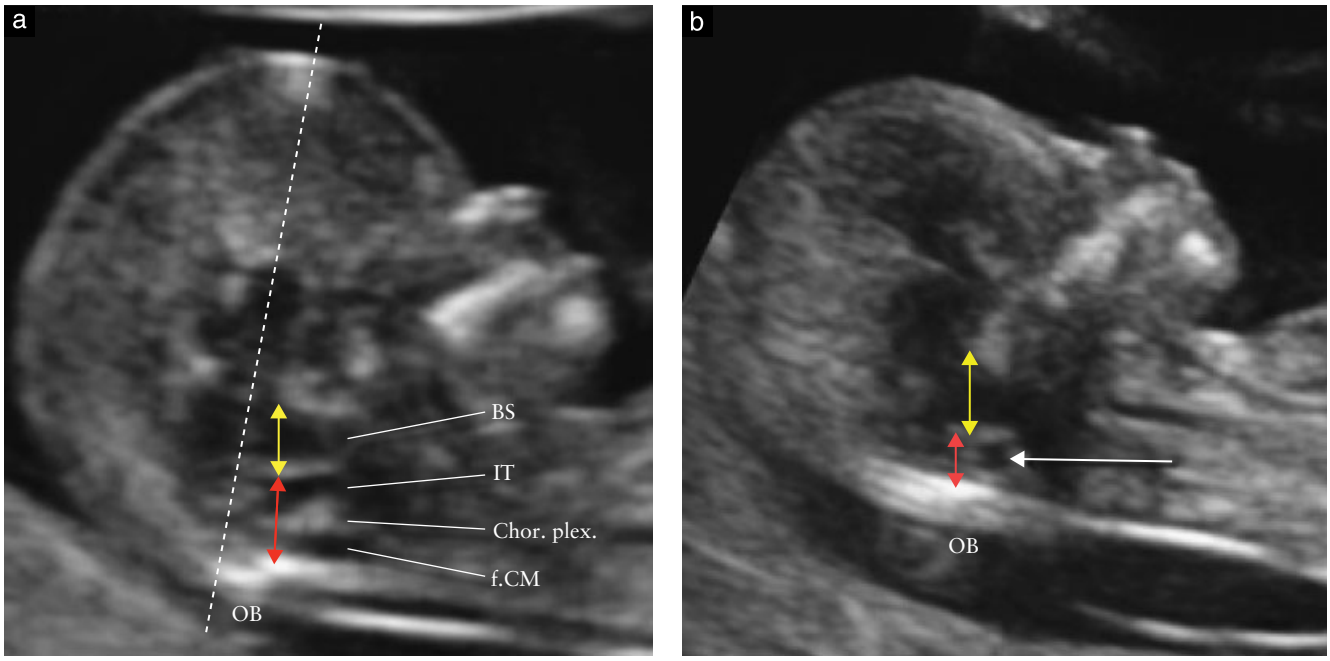

Figure 1 Posterior brain region in mid-sagittal view in a normal fetus (a) and in a fetus with spina bifida (b). In (a) the normal posterior brain anatomy is easily discernible: the intracranial translucency (IT) can be identified as fluid between the brainstem (BS) anteriorly and the choroid plexus (Chor. plex.) of the fourth ventricle posteriorly. The brainstem has a small diameter (yellow arrow) which is shorter than the brainstem–occipital bone (OB) distance (red arrow). Dashed line shows the plane seen in Figure 2a. f.CM, future cisterna magna. In (b) some fluid is still present (white arrow) but the landmarks of a normal IT are absent. No choroid plexus and no future cisterna magna are found. Common abnormal signs of an open spina bifida are present: the brainstem is thickened (yellow arrow) and the ratio of the brainstem to the brainstem–occipital bone (OB) distance (red arrow) is increased (> 1).

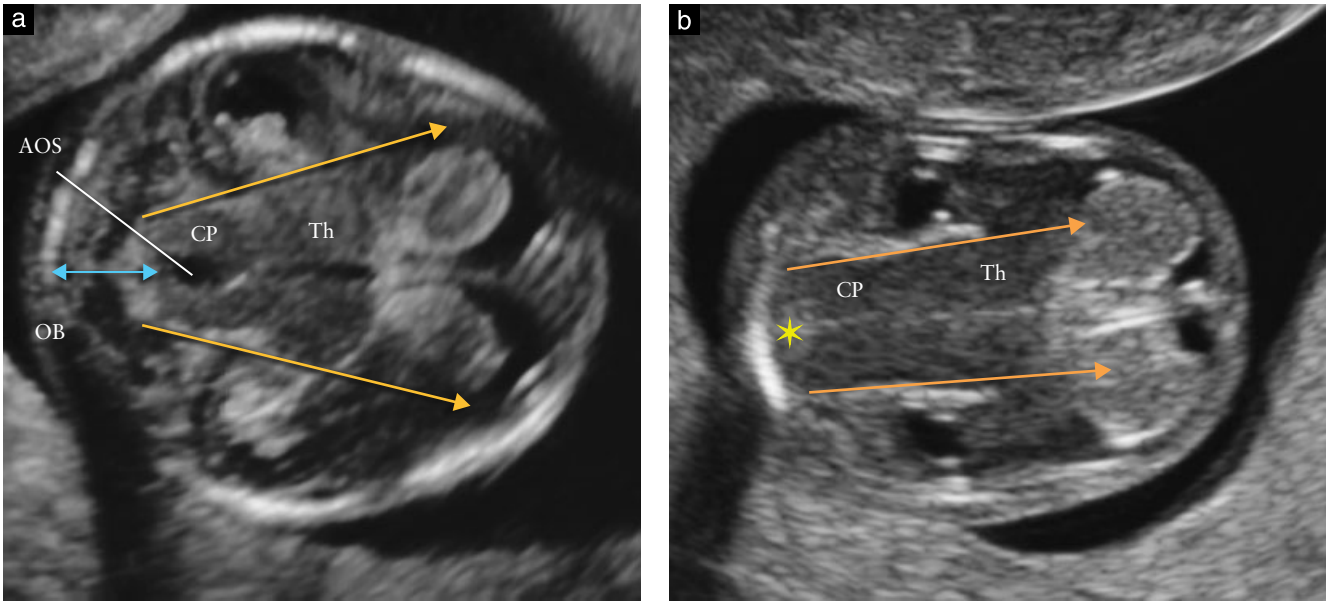

Figure 2 Transvaginal visualization of the posterior brain region in oblique axial view in a normal fetus (a) and in a fetus with spina bifida (b). (a) The plane demonstrated is along the dashed line shown in Figure 1a and is at the level of the diencephalon and midbrain, including both thalami (Th) with the fluid of the third ventricle in between and cerebral peduncles (CP) with the fluid of the aqueduct of Sylvius (AOS) in between. There is space between the AOS and the occipital bone (OB) (blue arrow) and the transition between cerebral peduncles and thalami forms an acute angle (orange arrows). (b) In comparison, in the fetus with spina bifida there is no space (yellow star) between the cerebral peduncles and occipital bone due to the posterior shift of the midbrain, and the transition between cerebral peduncles and thalami forms two parallel lines (orange arrows).

opinion, this can be achieved by visualizing the posterior brain region in the same mid-sagittal plane as is used in screening for aneuploidies. The feasibility of this approach of including examination of the posterior fossa in the 11–13-week screening scan has been demonstrated by the prospective detection of six cases of open spina bifida[7]. The mid-sagittal plane is more likely to be successful in comparison to additional new planes, such as the axial

oblique plane described below. Interestingly, analysis of the axial view of the head for indirect signs of spina bifida is not a new approach[16], but it has not translated over the last 10 years into better detection of spina bifida.

Axial view of the brain: useful in expert hands and with transvaginal ultrasound

Visualization of an axial plane of the head is part of the 11–13-week scan in many centers. It has three purposes: firstly, demonstration of a normal head shape; secondly, measurement of the biparietal diameter; and thirdly, exclusion of most cases of holoprosencephaly by demonstration of both choroid plexuses separated by the falx cerebri. However, for examination of the posterior fossa a more oblique plane is required[17].

Two papers in this issue of the Journal report on the axial view of the brain in fetuses with spina bifida[9,18]. In one, the authors used multiplanar three-dimensional ultrasound and compared the mid-sagittal and axial planes in normal fetuses and in fetuses with spina bifida[18]. While interesting, this approach is not particularly convenient in a routine clinical setting. In the other paper, the authors focused on the aqueduct of Sylvius and the midbrain (Figure 2) visualized using transabdominal and transvaginal approaches[9]. In our experience, the axial view of the posterior fossa can be better visualized by transvaginal than by transabdominal ultrasound. The transvaginal route reveals many details but needs expertise in understanding the structures of interest. A transvaginal examination is, however, only performed occasionally at 11–13 weeks and cannot be considered as a routine tool for screening.

An interesting observation regarding axial views of the posterior brain in cases of spina bifida was reported 10 years ago by Buisson *et al.*[16] and is reported again by Finn *et al.*[9] in this issue. In 2002, Buisson *et al.* described the appearance of the posterior brain in two fetuses with spina bifida and showed that, 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 (Figure 2b). Under normal conditions, transition from the cerebral peduncles to the thalami follows an acute angle (Figure 2a). Finn *et al.* also observed juxtaposition of the midbrain to the occipital bone, and suggested quantification of this observation by measuring the distance between the aqueduct of Sylvius and the occipital bone, which was reduced in their nine fetuses with spina bifida.

We believe that use of the axial plane of the posterior brain to identify parallelism of the cerebral peduncles and posterior shifting of the midbrain and aqueduct of Sylvius is not likely to be helpful for screening. This plane could, however, be useful to the specialist, especially when the mid-sagittal view reveals a suspicious finding. On transvaginal examination, when the mid-sagittal plane cannot be obtained easily, an axial view of the posterior region can provide enough information to distinguish a normal from a suspicious finding.

Conclusion

A screening test should be easy to perform and require little additional effort over that required for routine examination. 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. In the second trimester, routine examination of the fetal brain should include axial planes for measurement of the biparietal diameter, head circumference and cerebellar transverse diameter[19]. In cases with suspicious findings, comprehensive fetal neurosonography involving acquisition of additional coronal and sagittal planes is recommended. At the 11–13-week scan, however, the mid-sagittal view of the face is the standard view obtained in every fetus and we therefore believe that this view should be used to visualize the posterior brain region. Suspicious cases can undergo additional assessment by visualization of axial planes, either transabdominally or transvaginally, and by targeted examination of the spine. Preliminary prospective reports of spina bifida detected using this screening approach are encouraging[7], but large prospective studies and data on sensitivity and specificity are still needed.

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