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Intracranial translucency as a sonographic marker for detecting open spina bifida at 11-13^{+6} weeks scan: Our experience

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Abstract

Aims and Objectives: The fourth ventricle, seen as intracranial translucency (IT) at 11–13^{+6} weeks, has been reported to be obliterated in cases of open spina bifida (OSB). Our aim was to assess its role in detecting OSB at 11–13^{+6} weeks. **Materials and Methods:** This prospective study was conducted at foetal medicine unit from January 2014 to June 2015. All women who underwent both first and mid-trimester scan in our unit were included in the study. IT was categorized as normal, obliterated or not clear. Spine was examined during both scans. **Results:** Totally, 341 cases were included in the study. IT was found to be obliterated in four cases and not clear in one case. There was demonstrable OSB at 11–13^{+6} weeks in two cases, at 15–16 weeks in two cases and at 24 weeks in one case. In the remaining 336 cases with normal IT, spine was found to be normal at target scan. **Conclusion:** Mid-sagittal view of face that is routinely used to measure nuchal translucency (NT) can also be used to detect OSB. It is feasible to integrate IT into the routine 11–13^{+6} weeks scan.

Key words: First trimester; intracranial translucency; open spina bifida

Introduction

Neural tube defect (NTD) is a serious congenital anomaly associated with mortality and lifelong disability, making prevention of open neural tube defect (ONTD) a global problem.[1-5] Diagnosis of open spina bifida (OSB) in the antenatal period is routinely done in the mid-trimester using lemon and banana signs on ultrasound. Fourth ventricle is seen as intracranial translucency (IT) between 11 and 13^{+6} weeks of gestation. Obliterated IT has been identified as a marker for ONTD.[6,7] The objective of this study was to assess the role of IT for detecting OSB in the first trimester.

Materials and Methods

This is a prospective study conducted at foetal medicine unit from January 2014 to June 2015. The study group included all the women who underwent both first trimester (11–13^{+6} weeks) scan and mid-trimester scan in our unit.

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NT was measured in the mid-sagittal section of fetal head according to the Fetal Medicine Foundation (FMF), UK criteria by FMF-certified sonologists.[8-10]

FMF protocol for measurement of NT:

1. The gestational period must be 11–13 weeks and six days.
2. The fetal crown-rump length (CRL) should be between 45 and 84 mm.
3. The magnification of the image should be such that the fetal head and thorax occupy the whole screen.
4. A mid-sagittal view of the face should be obtained. This is defined by the presence of the echogenic tip of the nose and rectangular shape of the palate anteriorly, the translucent diencephalon in the center and the nuchal membrane posteriorly. Minor deviations from the exact midline plane would cause nonvisualization of the tip of the nose and visibility of the zygomatic process of the maxilla.
5. The fetus should be in a neutral position, with the head in line with the spine. When the fetal neck is hyperextended, the measurement can be falsely increased and when the neck is flexed, the measurement can be falsely decreased.
6. Care must be taken to distinguish between fetal skin and amnion.
7. The widest part of translucency must always be measured.

IT was identified in the mid-sagittal image of fetal face as a clear space bounded anteriorly by posterior border of brainstem and posteriorly by choroid plexus of fourth ventricle [Figure 1]. IT was categorized as normal, obliterated [Figure 2] or not clear [Figure 3]. Fetal spine was examined in detail in both first and mid-trimester scan.

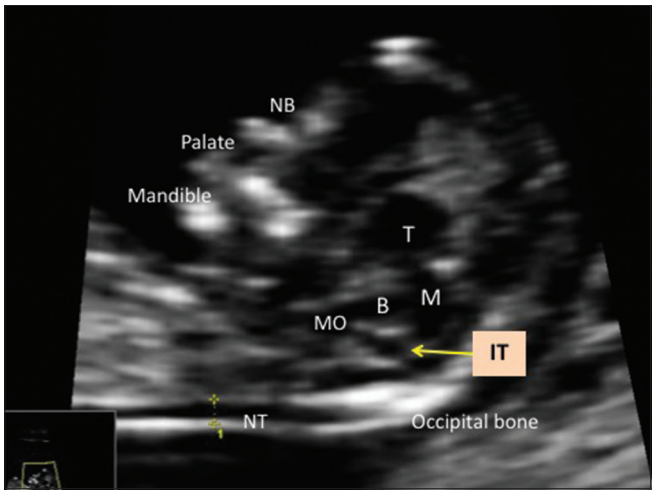

Figure 1: Ultrasound image of mid-sagittal section of fetal head showing normal IT. Thalamus (T), Midbrain (M), Brainstem (B), Medulla Oblongata (MO), Nuchal translucency (NT), and Nasal bone (NB)

Results

A total of 4345 scans were performed in our department during the study period. Of these, 640 women underwent scan between 11 and 13^{+6} weeks for combined first trimester screening (screening for aneuploidy using NT and double markers.) Out of 640 cases, 341 women who underwent mid-trimester scan in our unit were included in the study. IT was found to be obliterated in four cases, not clear in one and normal in 336 cases. In two out of four cases with obliterated IT, there was demonstrable OSB on the first trimester scan [Figure 4A and B] and suspicion of spina bifida in rest of the two [Figure 5]. The two cases with suspicion of spina bifida in the first trimester were reviewed at 16 weeks and OSB was demonstrated subsequently [Figure 6]. One case where IT was not clear due to high body mass index and previous cesarean scar and spine appeared to be normal in first trimester [Figure 7]. OSB was diagnosed at 24 weeks as patient reported to us late [Figure 8]. All the cases of OSB diagnosed either in first trimester or in mid-trimester were opted for termination of pregnancy. In 336 cases where IT was normal, spine was imaged in sagittal, coronal and transverse sections in mid-trimester target scan and was found to be normal [Figure 9]. All these cases delivered with us had normal postnatal outcome.

Discussion

NTD is associated with significant morbidity and mortality, which in turn has an impact on the emotional and financial status of the affected individual, families, health system and society.[2,5,11,12] Prevalence of ONTDs is found to be more in low socioeconomic countries as compared to middle and high socioeconomic countries.[13] Significant reduction in the prevalence of NTDs was noted

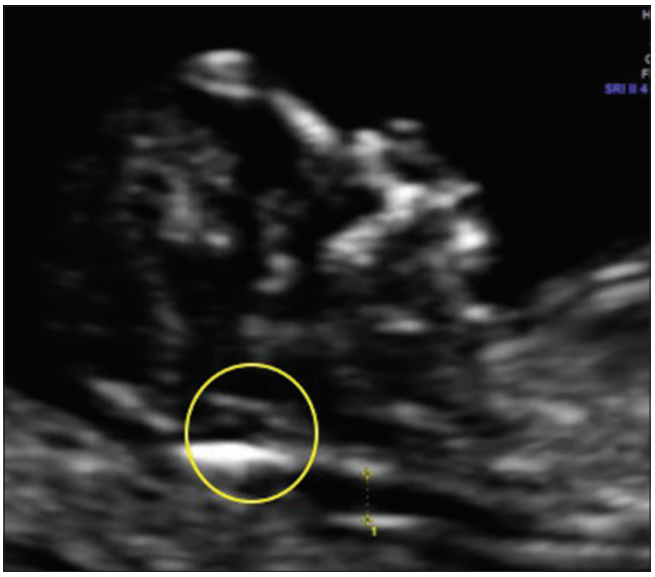

Figure 2: Ultrasound image of mid-sagittal section of fetal head showing obliterated IT

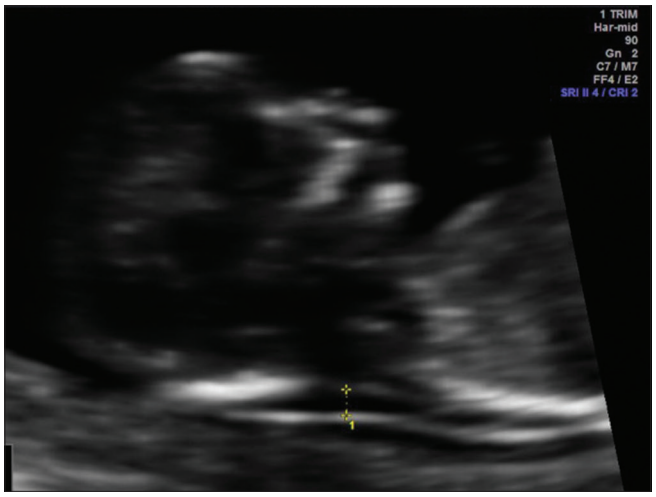

Figure 3: Ultrasound image of mid-sagittal section of fetal head where IT is not clear

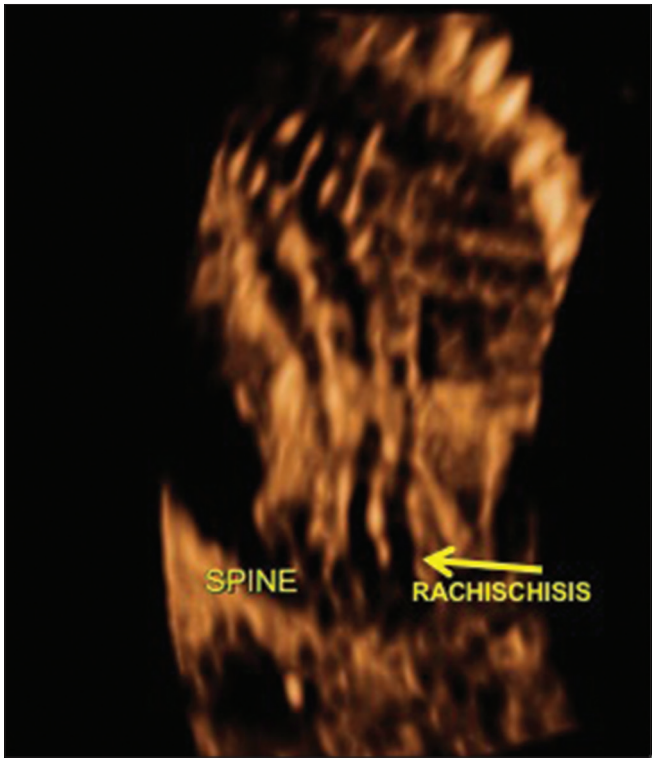

Figure 5: Ultrasound image of coronal section of fetal spine with suspicious NTD in the first trimester where IT was obliterated

in the countries where food fortification with folic acid was made as a mandatory program and this is being used as primary prevention in many countries (CDC).[12,14,15] India falls into the group of countries where the prevalence of NTD is high and there is no implementation of mandatory fortification of food with folic acid. Hence, secondary prevention of ONTD plays an important role to decrease the burden of ONTD.

In our practice, we have observed that the majority of OSB cases are diagnosed in the mid and late

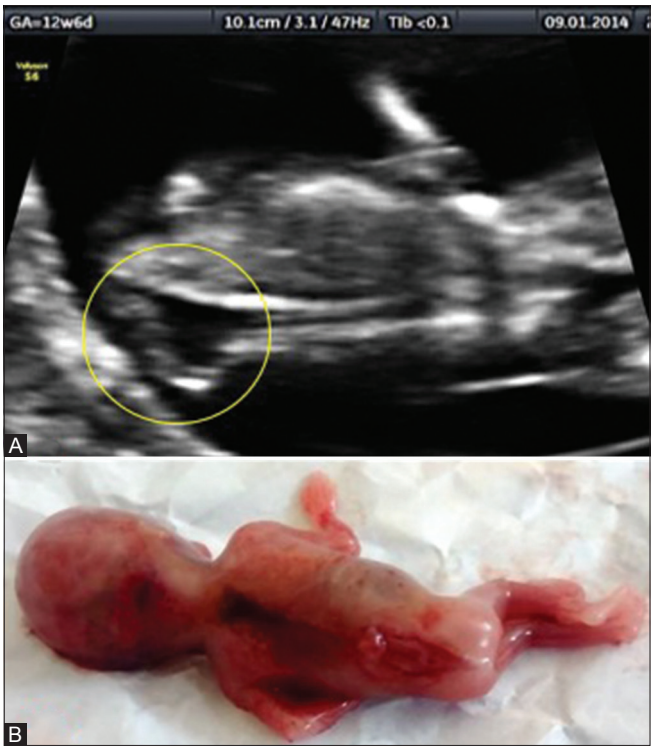

Figure 4 (A and B): (A) Ultrasound image of sagittal section of fetal spine showing ONTD in the first trimester where IT was obliterated. (B) Post termination picture of abortus for the same case

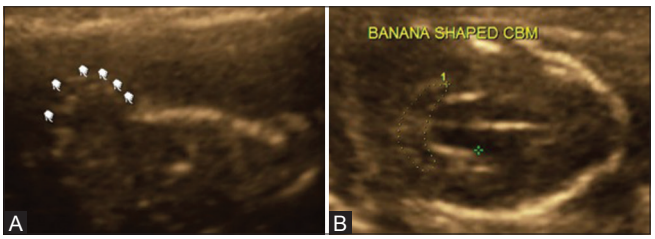

Figure 6 (A and B): (A) Ultrasound image of sagittal section of fetal spine at 16 weeks showing ONTD (pointer marked). (B) Axial section of fetal head at 16 weeks showing banana-shaped cerebellum (CBM)

mid-trimester [Figure 10]. Diagnosis of spina bifida in the mid-trimester is routinely done by direct visualization of the spinal defect or by identifying deformed cerebellum and obliterated cisterna magna as a "Banana sign" or frontal bossing as a "Lemon sign," which has almost 100% detection rate.[16,17] These signs are proposed to occur due to the caudal displacement of cerebellum as a result of leakage of cerebrospinal fluid. Similar hypothesis when extrapolated to first trimester is thought to be the reason for obliterated IT.

With widespread use of NT measurement for aneuploidy screening and detailed anatomical survey during 11 to 13^{+6} weeks of gestation, first trimester scan has become standard pregnancy care for the past two decades. In normal fetuses, the fourth ventricle is always visible as IT in the same plane of NT measurement and nasal bone evaluation. The

Figure 7: Ultrasound image of normal fetal spine in the case where IT was not clear

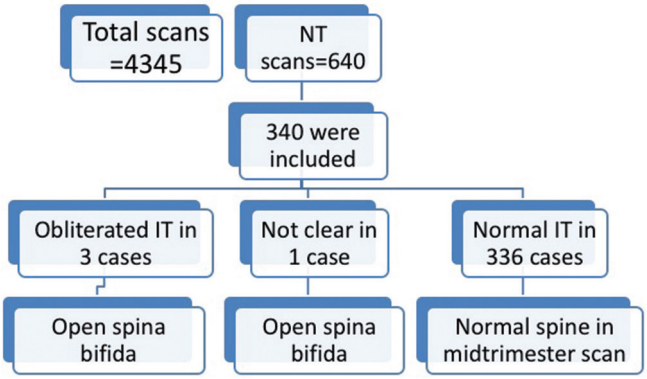

Figure 9: Flowchart explaining the results

median antero-posterior diameter increased from 1.5 mm at CRL of 45 mm to 2.5 mm at a CRL 84 mm. Obliterated IT as a marker for OSB at 11–14 weeks was first suggested by Rabi Chaoiu *et al.* and found that they had 200 normal cases with demonstrable IT and four OSB cases with obliterated IT.[6,7]

Our objective was to look at the feasibility of IT imaging during 11–13^{+6} weeks scan and to assess its role in early detection of OSB in the first trimester. This was a prospective study conducted at two fetal medicine centers and the study group included a total of 341 cases that came for NT scans. Out of 341, IT was clearly visualized in 336. When IT was present, spine was normal and OSB was excluded during the target scan, resulting in 100% specificity comparable to 99% specificity reported by Fong *et al.*[18]

All four with obliterated IT were confirmed with OSB either during first trimester or during 16 weeks scans. One case where IT was not clear due to maternal habitus had OSB on subsequent scan. This resulted in sensitivity of nonvisualization of IT for detection of OSB is 80% when compared to 50% by Fong *et al.*[18] The reason for better sensitivity in our study could be due to prospective nature of our study, which was exclusively done for evaluation of IT fulfilling all the criteria for it. There was no association

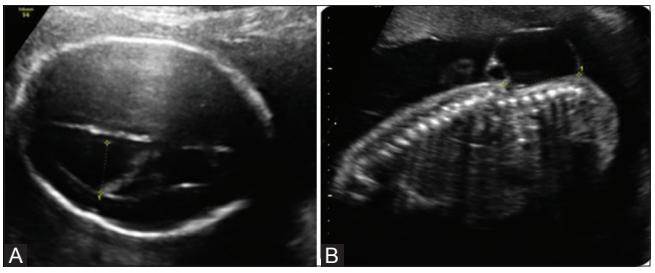

Figure 8 (A and B): (A) Ultrasound image of axial section of fetal head at 24 weeks showing ventriculomegaly. (B) Ultrasound image of sagittal section of fetal spine showing meningocele

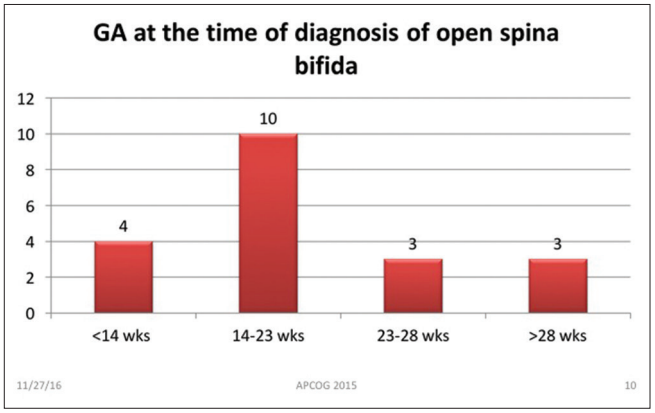

Figure 10: Bar diagram showing the relations of gestational age (GA) time of the diagnosis of OSB

between IT visibility and gestational age or CRL of the fetus. However, maternal habitus, previous scar has impact on visualization of IT and woman with high body mass index may pose difficulty in visualization of IT.

Conclusion

We found that IT is a reliable marker that can be easily evaluated in the same plane of NT measurement, which is routinely performed for first trimester aneuploidy screening. Obliterated IT has reasonable sensitivity for detection of OSB. These cases can be reviewed early for further evaluation of spine. Detection of spinal defect early in gestation facilitates safer method of termination of pregnancy. Presence of normal IT excludes OSB. The diagnostic performance of IT for detection of OSB is directly proportional to training and experience of the sonographer.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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