

A retrospective review of sonographic markers for detection of open spina bifida at 11-14 weeks

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Objective

The aim of this study was to review diagnostic and screening markers for open spina bifida (OSB) during first-trimester ultrasound scans.

Methods

We searched our database between the years 2014 and 2018 to identify cases of isolated OSB diagnosed at the first or second trimester ultrasound examination. This was a retrospective study of ultrasound images of fetal head obtained in the first trimester. We analyzed the sagittal planes, obtained for measurement of crown-rump length and nuchal translucency thickness, and the transverse planes, obtained for measurement of the biparietal diameter. The systematic review for the presence of all published ultrasonographic markers for OSB at 11- 14 weeks was performed, namely for intracranial translucency (IT), brain stem diameter (BS), cisterna magna (CM) and maxillo-occipital line in the mid-sagittal plane. Furthermore, other markers as crash sign, BPD diameter, dry brain and the aqueduct of Sylvius in the axial plane were evaluated.

Results

The markers of OSB were present in 25 out of 31 confirmed cases of spina bifida. IT was found to be obliterated in 6 cases. Reduced or absent CM was identified in 2 cases of OSB. The maxillo-occipital line is a straight line drawn along the superior border of the maxilla that touches the occipital bone posteriorly. The junction of the thalamus with the midbrain was reported to be above this maxillo-occipital line in normal fetuses and below this line in the fetuses with OSB. In our study this marker was found in 13 cases. Biparietal diameter was below the 5th centile in 13 cases. In 9 cases dry brain (decreased area of the lateral ventricles) was present. Crash sign and the aqueduct of Sylvius were not seen in our series, because the midbrain is visualized slightly caudal to the plane in which the biparietal diameter is measured. In 4 cases the ultrasound examination of fetal spine directly revealed the open spina bifida. No markers of OSB were found in 6 fetuses.

Conclusion

Several markers for early detection of fetuses with spina bifida have been recently proposed. Our results suggest that in particular combination markers using a morphometric approach (BPD diameter, IT measurement) with anatomical analysis of the posterior brain (identification and counting 3 spaces - BS, IT, CM and crash sign) may be a useful strategy in the first-trimester detection of spina bifida. A prospective evaluation of all these markers used independently or in a combined screening strategy is needed.