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PRACE KAZUISTYCZNE
położnictwo

Combination of intracranial translucency and 3D sonography in the first trimester diagnosis of neural tube defects: case report and review of literature

Kombinacja wewnątrzczaszkowej przezierności i ultrasonografii 3D w diagnostyce wad cewy nerwowej w pierwszym trymestrze ciąży: opis przypadku i przegląd literatury

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Abstract

Neural tube defects are congenital defects of the central nervous system caused by lack of neural tube closure. First trimester screening for aneuploidy has become widespread in the recent years. Fetal intracranial translucency (IT) can be easily observed in normal fetuses in the mid-sagittal plane. The absence of IT should be an important factor taken into consideration in the early diagnosis of open spinal defects. 3D ultrasonography is especially useful in cases of spinal anomalies where the visualization of the fetal structure is insufficient due to fetal position. We present a combination of intracranial translucency and 3D sonography used in the first trimester diagnosis of a neural tube defect case.

Key words: **neural tube defects** / **first trimester diagnosis** / **3D sonography** / / **intracranial translucency** /

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Strzeszczenie

Wady cewy nerwowej są wrodzonymi defektami centralnego układu nerwowego spowodowanymi brakiem zamknięcia cewy nerwowej. Badania skriningowe w pierwszym trymestrze w kierunku aneuploidii stały się w ostatnich latach bardzo rozpowszechnione. Płodowa przezierność wewnątrzczaszkowa może być oceniona w prawidłowych płodach w płaszczyźnie pośrodkowej. Brak przezierności wewnątrzczaszkowej (IT) powinien być istotnym czynnikiem ryzyka branym pod uwagę we wczesnej diagnostyce otwartych wad cewy nerwowej. Ultrasonografia 3D jest szczególnie przydatna w przypadkach gdy uwidocznienie struktur płodu jest niewystarczające z uwagi na pozycję płodu. Przedstawiamy kombinację przezierności wewnątrzczaszkowej i ultrasonografii 3D w diagnostyce wad cewy nerwowej w pierwszym trymetrze ciąży.

Słowa kluczowe: **wady cewy nerwowej** / **pierwszy trymestr** / **diagnostyka** / / **ultrasonografia 3D** / **przezierność przezczaszkowa** /

Introduction

Neural tube defects are congenital defects of the central nervous system caused by lack of neural tube closure [1]. Detailed 2D ultrasonography is needed if maternal AFP is above the cut-off value. Significant development in the ultrasound technology has enabled us to achieve better visualization of fetal structures. Three-dimensional (3D) ultrasonography, 3D computed tomography (3D-CT) and magnetic resonance imaging (MRI) are the complementary visualization techniques. First trimester screening for aneuploidy has become widespread in recent years [2].

We present a combination of intracranial translucency and 3D sonography in a first-trimester diagnosis of a case with neural tube defect.

Case report

A 29 year-old patient (gravida 1, para 0), at 11 weeks 6 days of gestation had a routine first trimester aneuploidy scan, as recommended by the Fetal Medicine Foundation for nuchal translucency and nasal bone. Crown-rump length (CRL), 52 mm, was compatible with the gestational age. NT was 1.6 mm. Nasal bone was normal. At the same mid-sagittal plane, posterior fossa was evaluated. The 4th ventricle was examined and intracranial translucency corresponding to the 4th ventricle could not be seen. Frontomaxillary facial angle was measured and found to be 57° (Figure 1).

After these investigations the spinal cord defect was also revealed by 3D sonographic examination (Figure 2).

The parents were immediately informed about the findings. They preferred to wait until the next sonographic examination (at 15-16 weeks of gestation) before deciding on pregnancy termination. In the second examination at 15-16 weeks of gestation, the spinal defect and secondary cranial findings such as lemon and banana signs were also observed. Postmortem examination was consistent with the prenatal diagnosis (Figure 3).

Maternal medical history was unremarkable and did not include any risk factors such as folic acid deficiency or others.

Discussion

Neural tube defects are classified as open and closed spina bifida by Tortori-Donati et al [3]. In open spina bifida, which is commonly associated with Chiari type 2 malformations, neural

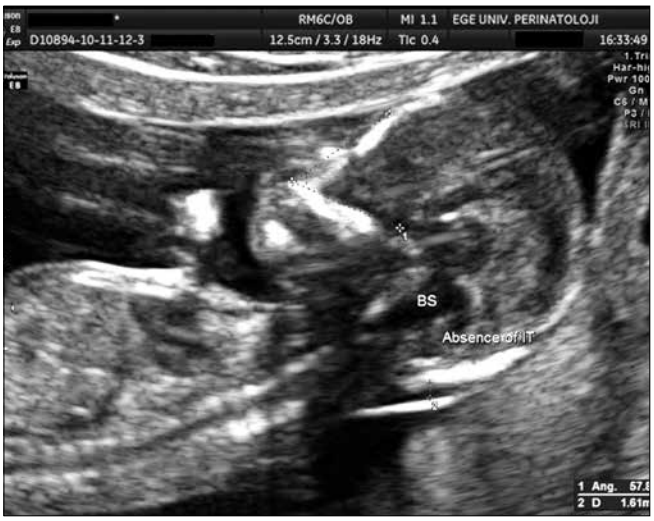

Figure 1. First trimester scanning of intracranial translucency, nuchal translucency and frontomaxillary angle in the same section.

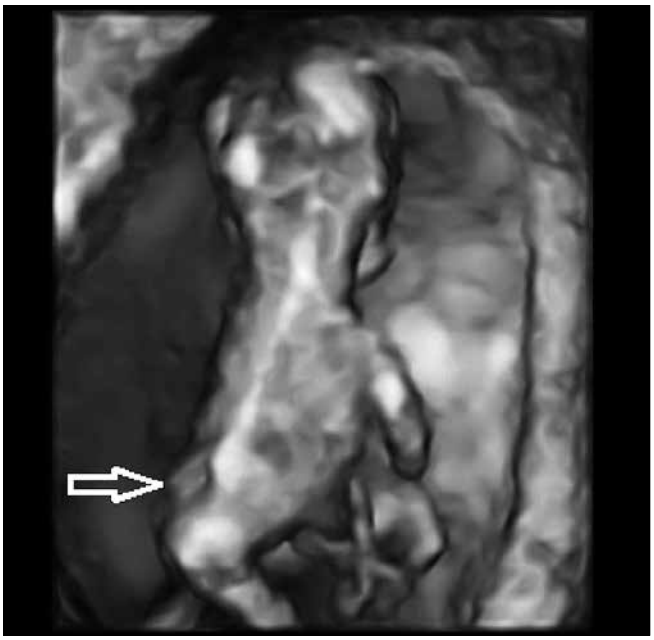

Figure 2. 3D image of a fetus with spina bifida (yellow arrow).

Figure 3. Postmortem examination of the same neural tube defect (yellow arrow).

elements and/or membranes protrude through a bone defect without skin covering [3-4]. Vertebral defect is enclosed by the skin in the closed forms and is rarely associated with Chiari type 2 malformations. Cranial anatomy is usually normal in closed forms and may be associated with subcutaneous cyst or mass. Prognosis is better in the closed type, usually asymptomatic in the affected individuals [3-4].

Measuring the maternal AFP level is an effective screening test for open spina. Next stage is a detailed 2D ultrasonography if maternal APF is higher than 2.5 MoM. In open spina bifida the leakage of cerebrospinal fluid to the amniotic cavity causes hypotension in the subarachnoid space and initiates the caudal displacement of brain. 'Banana' and 'lemon' signs are the cranial manifestations of open spina bifida in the second trimester scan. The first trimester appearance is manifested by loss of intracranial translucency due to the compression on the 4th ventricle caused by the displacement of hindbrain caudally [5]. In the closed forms of spina bifida AFP do not increase both in maternal serum and amniotic fluid and cranial anatomy is ubiquitously normal [4-5].

Recently, in many European countries the first trimester screening for aneuploidy has become very common and is performed in all pregnant women. Nuchal translucency and nasal bone are assessed in the mid-sagittal plane, in accordance with the recommendations of FMF. Posterior fossa can be observed with nasal bone and NT synchronously. The fourth ventricle, BS diameter, BSOS diameter and BS/BSOS are examined in the evaluation of the posterior fossa. In normal fetuses, the 4th ventricle presents as intracranial translucency parallel to NT [5-6]. The measurement is similar to the NT: anterior-posterior diameter of IT is measured [5-6]. Anterior border is the posterior border of the brain stem and posterior border is the choroid plexus of the fourth ventricle [5-7]. In open spina bifida IT may be absent.

Chaoui at al., performed a retrospective investigation on IT at 11-13 weeks gestation scans in a group of patients including 200 normal fetuses and 4 fetuses with spina bifida. In normal fetuses the fourth ventricle was visible in all cases but in the four fetuses with spina bifida, the fourth ventricle was compressed with the displaced hindbrain and IT could not be viewed in all four cases [6].

3D ultrasonography is especially useful in spinal anomalies where the visualization of fetal structure is insufficient due to fetal position [2]. Also, images should be assessed and manipulated later with 3D technology. Other complementary methods include MRI and 3D CT scanning. MRI may be used for further assessment of fetal abnormalities. In addition, 3D CT scanning offers a different way of imaging and is performed in the third trimester.

In conclusion, the preliminary studies mentioned above and our case report demonstrate that at 11-13 weeks gestation intracranial translucency can be easily observed in normal fetuses in the mid-sagittal plane. The absence of IT should be an important factor taken into consideration in the early diagnosis of open spinal defects.

Declaration of interest statement:
The authors declare no conflicts of interest.

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