Prenatal diagnosis of Dandy–Walker syndrome in early pregnancy presenting with increased nuchal translucency and generalized edema at 13 weeks of gestation

Prenatal diagnosis of Dandy–Walker Syndrome (DWS) is very crucial to fetal well-being. DWS has been visualized by prenatal ultrasound before 20 weeks of gestation, even as early as 13 weeks of gestation (Sherer et al., 2001). Nonetheless, to the best of our knowledge, only five cases of DWS presenting with increased nuchal translucency (NT) have been reported (Souka et al., 1998, 2001). We recently encountered a fetus presenting with an NT of 7.5 mm and generalized edema on a routine ultrasound scan at 13 weeks of gestation. A preamniocentesis ultrasound scanning illustrated DWS at 16 weeks of gestation. Karyotyping by amniocentesis turned out to be a normal male, 46,XY. The autopsy examination revealed DWS only, without any other abnormalities.

A 25-year-old woman, gravida 2, para 1, came to our prenatal clinic at 13 weeks of gestation for her first prenatal visit. A review of her past obstetric history revealed that she had given birth to a normal male fetus during her last pregnancy and no past history or family history was remarkable. At our clinic, we performed a routine prenatal ultrasound scan for assessing the basic biometry as well as the NT. The scan revealed a fetus with a crown-rump length (CRL) of 6.7 cm, with an increased NT, 7.5 mm in thickness and generalized edema (Figure 1A). As no other abnormality was noted at that time, an amniocentesis was scheduled at 16 weeks of gestation.

At 16 weeks of gestation, another ultrasound scan was performed before amniocentesis. The NT and generalized edema had disappeared but dilated lateral ventricles and a large cyst of the cisterna magna and a vermis defect were seen (Figure 1B). Prenatal diagnosis of DWS was made and amniocentesis was performed for karyotyping. The karyotyping turned out to be a normal male, 46,XY.

After genetic counseling, the couple chose to terminate the pregnancy. The fetus was aborted 10 h after misoprostol 200 mg was applied to the internal os. The fetus was sent for autopsy after abortion. Autopsy examination showed agenesis of the cerebellar vermis, ependymal cyst in the fourth ventricle, dilated lateral as well as third and fourth ventricles. The autopsy confirmed prenatal diagnosis of DWS. Under autopsy examination, the other organs and systems, including cardiovascular, gastrointestinal and musculoskeletal systems, were normal.

DWS was characterized by the association of ventriculomegaly of a variable degree with a large cisterna magna plus a defect in the cerebellar vermis (Pilu et al., 2000). Until now, prenatal ultrasound has been the cornerstone of prenatal diagnosis of DWS. Although the karyotyping in our case was normal and the other structures besides the central nervous system were also intact, the complete

References


vermis defect, the cystic cisterna magna as well as the four dilated ventricles indicated a very poor prognosis.

In conclusion, performing an ultrasound scan at 10 to 14 weeks of gestation may help detect abnormal NT, which is often combined with at least 30 kinds of fetal organic defects. NT abnormality also further alerts the obstetricians to perform a systemic fetal ultrasound for detecting any subtle structural abnormalities in utero. All these advantages may contribute to increasing our knowledge in maternal–fetal medicine and provide vital information for genetic counseling.

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REFERENCES


Prenatal diagnosis of right pulmonary agenesis associated with VACTERL sequence

A 30-year-old primigravida woman was referred to the hospital at 30 weeks’ gestation because of suspected fetal pulmonary abnormality. She and her husband were non-consanguineous and healthy. There was no family history of congenital malformations. She did not have teratogenic medication, recent infection, diabetes mellitus, or hypertension during this pregnancy. Obstetric ultrasound revealed a single live fetus with a biparietal diameter of 7.6 cm (equivalent to 30 weeks’ gestation), an abdominal circumference of 22.3 cm (equivalent to 26 weeks’ gestation), a femur length of 5.7 cm (equivalent to 29 weeks’ gestation), a normal amniotic fluid index of 10.6 cm, and a mediastinal shift towards the right side with the right part of the heart contiguous with the ribs. Normal lung tissue was observed on the left side, while no parenchymatous or cystic tissue was present on the right side (Figure 1). The colour Doppler sonography showed dilated pulmonary trunk, right atrial dilatation, right ventricular hypertrophy, and absence of a right pulmonary artery. The findings were consistent with the diagnosis of right pulmonary agenesis (Figure 1). The stomach, liver, and spleen were normally positioned. The pregnancy progressed uneventfully, and the woman delivered a 3220-g male baby at term. The Apgar scores were 4 and 6 at 1 and 5 min respectively. Chest X-ray demonstrated absence of the right lung and fused vertebrae at the thoracic level. Multiple anomalies including cleft palate, an imperforate anus,

**Figure 1**—Prenatal ultrasonography of the fetus. (A) The 13-weeks’ scan revealed an increased nuchal translucency (7.5 mm). The crown-rump length (CRL) was 6.7 cm. The white arrow indicates the amniotic membrane. (B) The 16-weeks’ scan depicted a complete vermis defect (VD, small white arrow), a cyst of the cisterna magna (large white arrow) and ventricular dilatation (arrow head).