A False-Positive Diagnosis of a Prenatal Encephalocele on Transvaginal Ultrasonography

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The ability to correctly diagnose an encephalocele is important for both the management and outcome of a pregnancy. Not only has diagnostic evaluation with transvaginal ultrasonography been found to improve accuracy in the diagnosis of encephaloceles, but it has also helped in earlier diagnosis because of its high resolution and access to the fetal intracranial anatomy. It has been shown in previous case reports to correctly identify encephaloceles in cases in which the diagnosis was unable to be made on the basis of transabdominal ultrasonography.

In the case presented, transvaginal ultrasonography proved not to be diagnostically superior to abdominal ultrasonography for the diagnosis of an encephalocele. On the basis of transvaginal ultrasonography, an incorrect diagnosis of an encephalocele was made, which was then refuted by transabdominal ultrasonographic findings. In this case, it was fetal hair that was initially misinterpreted as an encephalocele on transvaginal ultrasonography.

Case Report

A 26-year-old woman, gravida 1, para 0, was referred to our perinatal center at 32.7 weeks' gestation for preterm labor. Cervical length assessment for preterm labor on transvaginal ultrasonography revealed what appeared to be a defect in the occipital skull as well as a suspected meningocele (Fig. 1). Because of the appearance and location of the anomaly, it was thought to be either an encephalocele or an occipital meningocele. This finding was not seen on previous ultrasonography done at 19 weeks' gestation, nor could it be duplicated by transabdominal ultrasonography at the time of diagnosis (Fig. 2).

Further analysis of the intracranial anatomy showed a normal cisterna magna, cerebellum, and third and lateral ventricular size. However, the head size did measure approximately 4 weeks behind what was expected by dates. An additional finding on transabdominal ultrasonography was abundant fetal hair in the occipital

Received December 12, 2000, from the Department of Obstetrics and Gynecology, Creighton University School of Medicine, Omaha, Nebraska. Revision requested January 17, 2001. Revised manuscript accepted for publication April 10, 2001.

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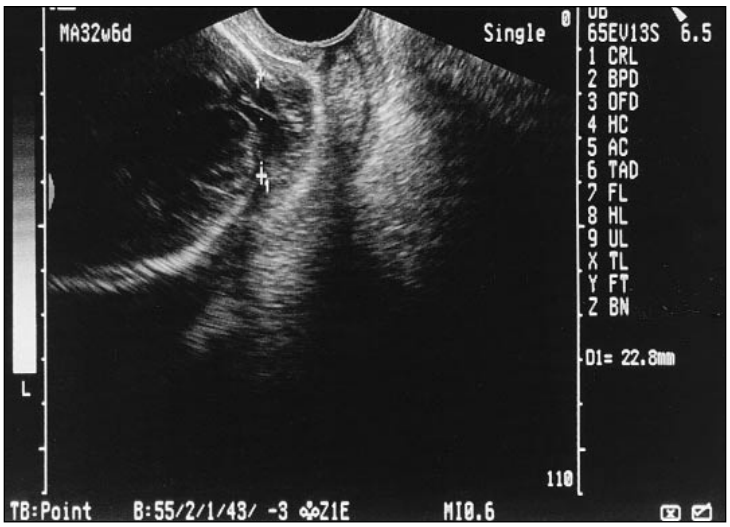


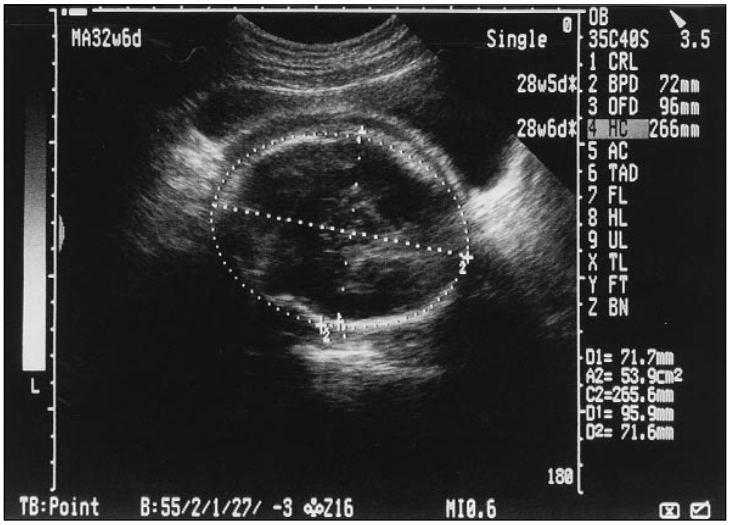
Figure 1. Transvaginal ultrasonography showing a break in the occipital skull with protrusion of a sac.

region. The remainder of the examination findings were normal, including the amniotic fluid volume. At birth, the cervical spine and occiput were intact. The infant did, however, have abundant hair in the occipital region.

Discussion

Incomplete closure of the neural tube and overlying mesoderm at the rostral end of the tube produce an encephalocele. Herniation of only the meninges through this bony defect defines a meningocele, whereas herniation of brain tissue through the defect defines an encephalocele. Encephaloceles are subdivided by the location of the bony defect and include occipital, parietal, and frontal, with occipital being the most common. The outcome for infants with encephaloceles is poor, with only a 50% chance of survival and a 74% prevalence of mental retardation. In contrast, the prognosis for meningoceles is much better, with no associated deaths and a 60% chance of normal mental development.[1] Conditions found to be associated with encephaloceles include rubella, diabetes, genetic syndromes, and amniotic band syndrome.

Figure 2. Transabdominal ultrasonography showing an intact occipital skull with no evidence of a protruding sac.



The differential diagnosis for an encephalocele includes a cystic hygroma, a teratoma, a branchiogenic cyst, a hemangioma, and scalp edema.[1] Misinterpretation of fetal hair as a meningocele, an encephalocele, or a cystic hygroma has also been reported. Two notable cases by Quinlan[2] described the misinterpretation of fetal hair in the occipital region as a craniocervical meningocele. In both cases the diagnosis was made in the third trimester, and both cases were complicated by polyhydramnios.[2] Diagnostic confusion in the presence of polyhydramnios or excessive fetal hair has been shown to occur because the ultrasonographic appearance of fetal hair becomes one of a continuous, thin-walled, fluctuant membrane in the occipital or suboccipital area. This membrane may then be misinterpreted as either a meningocele or an encephalocele, as occurred in the 2 cases reported by Quinlan[2] and the case reported here. It is important to note that fetal hair is not shown on ultrasonography until 24 weeks' gestation; therefore, diagnostic confusion should not occur before that time.[3]

Improved prenatal diagnosis of encephaloceles has been shown with transvaginal ultrasonography. Two cases have been reported in which transvaginal ultrasonography facilitated the correct diagnosis of encephaloceles when the correct diagnosis was not possible on the basis of transabdominal ultrasonography. In the first case, transabdominal ultrasonography at 17 weeks' gestation failed to show an encephalocele because the fetus was in a cephalic position deep in the pelvis. Evaluation with transvaginal ultrasonography allowed visualization of the fetal head, showing the presence of a frontal encephalocele.[4] In the second case, transabdominal ultrasonography at 12 weeks' gestation showed a possible occipital meningocele.

Confirmation with transvaginal ultrasonography revealed brain tissue in the sac, which changed the diagnosis to an encephalocele.[5] In both cases, the higher resolution and improved access to the fetal intracranial anatomy offered by transvaginal ultrasonography allowed for the correct diagnosis.

In the case presented here, third-trimester transvaginal ultrasonography led to a false-positive diagnosis of an encephalocele. Further analysis with transabdominal ultrasonography revealed that the initial bony defect was actually abundant fetal hair in the occipital region. In contrast to the cases described above, transvaginal ultrasonography did not prove to have diagnostic advantages over transabdominal ultrasonography later in pregnancy. Rather, it was the transabdominal examination that was crucial in making the correct diagnosis of misinterpreted abundant fetal hair. Therefore, it is important that the diagnosis of an encephalocele later in pregnancy be made with transabdominal ultrasonography and that the diagnosis of an encephalocele before the appearance of fetal hair be made with transvaginal ultrasonography. With either ultrasonographic modality, fetal hair should always be considered as part of the differential diagnosis of encephaloceles later in pregnancy.

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