



Occipital Encephalocele

Oksipital Ensefalosel

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Abstract

Encephalocele is a neural tube defect characterized by sac-like protrusions of the brain and the covering membranes through an opening in the skull. Encephalocele is less common than other neural tube defects. In this case we presented a 21 years old 20 weeks pregnant woman with fetal occipital encephalocele accompanying lemon sign, normal posterior fossa imaging and normal level of maternal serum alpha-fetoprotein (MSAFP).

Key words: Neural tube defect, encephalocele, lemon sign, maternal serum alpha-fetoprotein

Özet

Ensefalosel beyin dokusunun kafa tasındaki bir açıklıktan dışarıya çıkmasıdır. Ensefalosel diğer nöral tüp defektlerine kıyasla daha az yaygındır. Bizim vakada ultrason görüntülemesinde oksipital ensefalosele eşlik eden limon belirtisi ve normal bir posterior fossa izlenmekle birlikte, normal düzeyde maternal serum alfa-fetoproteini (MSAFP) olan 21 yaşındaki 20 haftalık gebe sunulmaktadır.

Anahtar kelimeler: Nöral tüp defekti, ensefalosel, limon belirtisi, maternal serum alfa-fetoprotein

Introduction

Encephalocele has a reported prevalence of 0,8 to 5 per 10,000 live births¹. The location of the defect is midoccipital in 80%, followed by the frontal midline in 13% of cases². The definitive diagnosis can be made only by visualizing the bone defect in addition to the hernial sac³. The MSAFP is frequently elevated⁴ but is not always so⁵. According to Guthkelch⁶, the mortality rate is 71% for infants with an encephalocele. When the findings are pronounced and when there are associated anomalies that are detected early before viability is reached, pregnancy termination may be considered⁷. Nicolaidis and associates

(1986) described frontal bone scalloping like the "lemon sign" in second-trimester fetuses with open spina bifida⁸. Although the exact pathogenesis is unknown, it has been postulated that the decrease in the intraspinal pressure in neonates with spina bifida causes the brain to shift downward. However, this theory does not explain why the lemon sign is present in fetuses with a normal posterior fossa. Therefore, an alternative theory has been proposed that the lemon sign might be due to a primary skeletal developmental disorder and that the contour of the skull is a result of mesenchymal dysplasia of the cranium⁹.

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Many centers now use specialized sonography as the primary method of evaluating an elevated MSAFP as recommended by the American College of Obstetricians and Gynecologists (2003). Women should be counseled regarding risks and benefits of both diagnostic tests, the risk associated with their degree of MSAFP elevation or with other risk factors, and the quality and findings of the sonographic examination before making a decision.

Fetal body wall defects uncovered by integument, such as neural tube defects and ventral wall defects, permit alpha-fetoprotein (AFP) to leak into the amnion fluid, resulting in dramatically increased MSAFP levels⁸.

We describe one case of closed defect with lemon sign diagnosed by ultrasound and normal MSAFP level at 20 weeks of gestation.

Case report

A 23-year-old multiparous woman was referred to our clinic for detailed ultrasound evaluation. During her first pregnancy, she had no fetal abnormality and delivered healthy baby. Maternal history did not reveal any chronic diseases, which could alter fetal development. From detailed obstetric history of current pregnancy, we figure out that pregnant was not exposed to any teratogenic medicaments, withal she did not used a folic acid contribution also. In her current pregnancy the ultrasound examination at 20 weeks 1 day showed appreciable widening of the normal cervical spinal echoes, and a well-defined 10x9 mm cystic area "attached" to the lower occipital area. The biparietal diameter of the fetal head and posterior fossa was within normal limits, lemon sign was detected, the ventricular

echoes appeared to be normal (Figure 1).



A



B

Figure 1. A Ultrasound image of occipital encephalocele B Ultrasound image of lemon sign

The MSAFP concentration was 22,4 ng/ml (0,99 MoM), within the normal range for 20 gestational weeks. In view of the conflicting evidence of a neural tube defect, the ultrasonic scan was repeated and the previous findings were confirmed. After detailed counselling, the pregnancy was terminated. Examination of the aborted fetus showed a completely skin-covered occipital encephalocele without concomitant macroscopic anomaly (Figure 2). Nevertheless, encephaloceles could be associated with triploidy, in our case, there was no abnormality in genetic evaluation of postabortal material.



A



B

Figure 2. A, B Occipital encephalocele

Discussion

Occipital encephalocele is a rare dysraphia with incidence varying according to geographic region. Over 10% of neural tube defects are covered by skin, the lesions ranging from small innocuous areas of spina bifida occulta to large myeloceles and encephaloceles, many of which result in major handicap.

Since the skin cover precludes the permeation of alpha-fetoprotein from the fetal cerebrospinal fluid into the amniotic cavity, MSAFP studies have been unhelpful in such cases¹⁰. Because MSAFP is insufficient in these kind of cases for early detection of encephalocele, ultrasound is the most preferable choice for diagnosis.

Regardless the fact that our case belongs to low-risk group for neural tube defects (normal level of MSAFP and non-complicated obstetric history), precise ultrasound evaluation gave us an opportunity to diagnose encephalocele in fetus. The lemon sign generally associated with spina bifida due to brain downward shifting that causing the obliteration of posterior fossa⁹. In our case, the closed defect of the neural tube abnormality was associated with lemon sign, without abnormality in posterior fossa. Our case support the opinion which postulates that compared with MSAFP performed alone for screening, routine second-trimester ultrasonography was more likely to discover an NTD.

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