What is your diagnosis?

A 32-year-old patient who was G9 P6 Y6 A2 was admitted to our clinic for detailed examination at the 21st week of gestation. Her medical history revealed that there was no antenatal follow-up. Abdominal sonographic examination showed one live fetus at 20 weeks of development. Detailed ultrasonographic examination revealed a 2x2 cm cystic enlargement localized in the fetal posterior fontanelle (Figure 1). Magnetic resonance imaging (MRI) was performed after the ultrasonography (Figure 2). What is your diagnosis?
Answer

This case shows the importance of fetal MRI in prenatal diagnosis. With only sonographic examination, the most probable diagnosis was encephalocele. But, with the addition of MRI, the diagnosis of this case changed. The lesion was localized under the skin and there was no relationship with intracranial structures. A cystic lesion was found in the parieto-occipital region with a size of 2x1x5x1 cm had a hypodense appearance on T1 and a hyperdense appearance on T2 (Figure 2). Intracranial structures were normal on MRI. With all these findings, the lesion was determined to be a subcutaneous epidermal cyst. The family was informed and it was decided to continue the pregnancy. However, the patient was lost to follow-up.

The use of fetal MRI was first reported in 1983 (1). Since then, with rapidly increasing technological developments and accumulated knowledge and experience, MRI is predominantly used for central nervous, respiratory and gastrointestinal anomalies because of the limitations of ultrasonography. MRI is not recommended in the first trimester due to a lack of sufficient information about its safety (2).

Encephalocele is an extremely rare neural tube defect. The incidence of encephalocele is 0.8-5/10,000 live births (3). Although the mechanism of encephalocele is not completely known, an anterior neural tube closure defect is the most likely mechanism (3). As a result of this fetal defect, changes occur to the calvarium and dura mater, along with herniation of the leptomeninges to the extracranial region. This sac sometimes consists of only cerebrospinal fluid and sometimes cerebrospinal fluid and brain tissue. It can appear in the occipital, frontal and basal regions. The localization of herniation is associated with a racial distribution. The occipital localization of encephalocele is the most recognizable type during the antenatal period. Because of the localization and appearance of the cyst, our initial diagnosis was encephalocele. However, MRI was also used for a differential diagnosis and with this method we could provide sufficient information to the family. Obesity and oligohydramnios have negative effects on ultrasonographic images, but these conditions have no negative effects on MRI. On the other hand, MRI has some limitations, like excessive fetal movement and fetal position changes. Today, images are obtained using an ultra-fast image capture technique (4). Before the procedure, an empty stomach and an empty bladder are recommended. Imaging is often performed in the supine position, but in later weeks of pregnancy, imaging is performed in the left lateral position because of supine hypotension.

Our case demonstrates once again that for soft tissue imaging, MRI is specific and sensitive. Our case also shows that T2 imaging is best for soft tissue while T1 imaging is more sensitive for hemorrhage, adipose tissue and diaphragmatic hernia.

References