Association of omphalocele and craniorachischisis totalis: The role of three-dimensional ultrasonography with diagnostic features

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Özet

Omfalosel ve kranioraşişiz totalis birlikteliği: Tanısal özellikleriyle birlikte üç boyutlu ultrasonografinin yeri

Omfalosel; karın içeriğinin ince bir membran içinde fıtıklaştığı karın ön duvarı defektlerindendir. Yüksek morbidite ve mortalite gösteren omfalosel en sık görülen karın ön duvarı defetlerindendir. İnsidansı yaklaşık olarak 5000 canlı doğumda 1'dir. Kranioraşişiz totalis; spinal defektle devamlılık gösteren anensefalidir. Defekt konsepsiyon sonrası 28 günlük sürede nöral tüpün kapanmaması ile oluşur ve fetal kayıp, ölü doğum ya da yenidoğan ölümüyle sonuçlanır. Prenatal tanısal araç iki anomali için de ultrasonografidir. Çoğu vakada iki-boyutlu konvansiyonal ultrasonografi kesin tanı için yeterli görüntüleme sağlar. Bu raporda omfalosel ve kranioraşişiz totalis birlikteliğini üç-boyutlu ultrasonografik ve post-abortal bulgularıyla sunuyoruz.

Anahtar Kelimeler: Omfalosel, Anensefali, Prenatal tanı, Karın ön duvarı defektleri, üç boyutlu görüntü.

Abstract

Omphalocele is an anterior abdominal wall defect with the herniation of the visceral contents covered by a thin membrane. It is one of the most common malformations of the anterior abdominal wall, with high morbidity and mortality. The incidence of omphalocele is nearly 1 in 5,000 live births. Cranirachischisis totalis is anencephaly with a contiguous spinal defect. The defect occurs with the failure of neural tube to close within 28 days after conception and leads to fetal loss, still birth, or neonatal death. Prenatal diagnostic tool for both anomalies is ultrasound. In most cases two-dimensional conventional ultrasound provides sufficient images for the accurate diagnosis. In this report, we present the association of omphalocele and craniorachischisis with the three-dimensional ultrasonographic and post-abortal findings.

Key Words: Omphalocele, Anencephaly, Prenatal diagnosis, Abdominal wall defects, 3D Image.

Introduction

Omphalocele is an anterior abdominal wall defect. It occurs at the base of the umbilical cord. Only the parietal peritoneum covers the herniated abdominal contents. In addition, it is one of the most common malformations of the anterior abdominal wall, with high morbidity and mortality. The incidence of omphalocele is nearly 1 in 5,000 live births. Mesoderm failure to replace the body stalk results the defect(1).

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Differential diagnosis of omphalocele and gastroschisis is important, because the higher rate of associated malformations with omphalocele, particularly aneuploidies, changes the management options. Fortunately, with the advances in high-resolution ultrasound (US) technology, the early and accurate diagnostic chance has been increased(2). Anencephaly is the absence of cranial vault and telencephalon with the coverage of the necrotic remnants of the brain stem and rhombencephalon by a vascular membrane. When the anencephaly is with the contiguous spinal defect, it is called "craniorachischisis totalis". The defect occurs as a result of the failure of neural tube to close within 28

days after conception and leads to fetal loss, still birth, or neonatal death(3-4).

Prenatal diagnostic tool for both anomalies is ultrasound. In most cases two dimensional conventional US provides sufficient images for the accurate diagnosis.

In this report, we will discuss the role of three-dimensional US in the diagnosis and presentation of a case of omphalocele with craniorachischisis totalis diagnosed in our department.

Case

A 22-year-old woman with her first pregnancy at the 17th week was referred to our centre with the diagnosis of fetal abdominal mass. Her medical history, family history and triple screening test were unremarkable. In two dimensional US an abdominal mass of 22.9 x 22.02 mm in diameters was observed. Two-dimensional US gave the image of a separate mass just above the umbilical cord insertion site. By using Doppler US and three-dimensional US, an omphalocele including the liver was identified. We performed a detailed US examination and identified anencephaly with the contiguous spinal defect. After the three-dimensional US demonstration of the case, the family decided to terminate the pregnancy. Three-dimensional US pictures were the main facts clearing the uncertainty of the family. We obtained written informed consents for pregnancy termination and publication of the case. A single dose of 200 mcg misoprostol administrated vaginally succeeded the complete abortion of the gestational product. Post-abortal examination confirmed the 3D US findings (Figure 1).

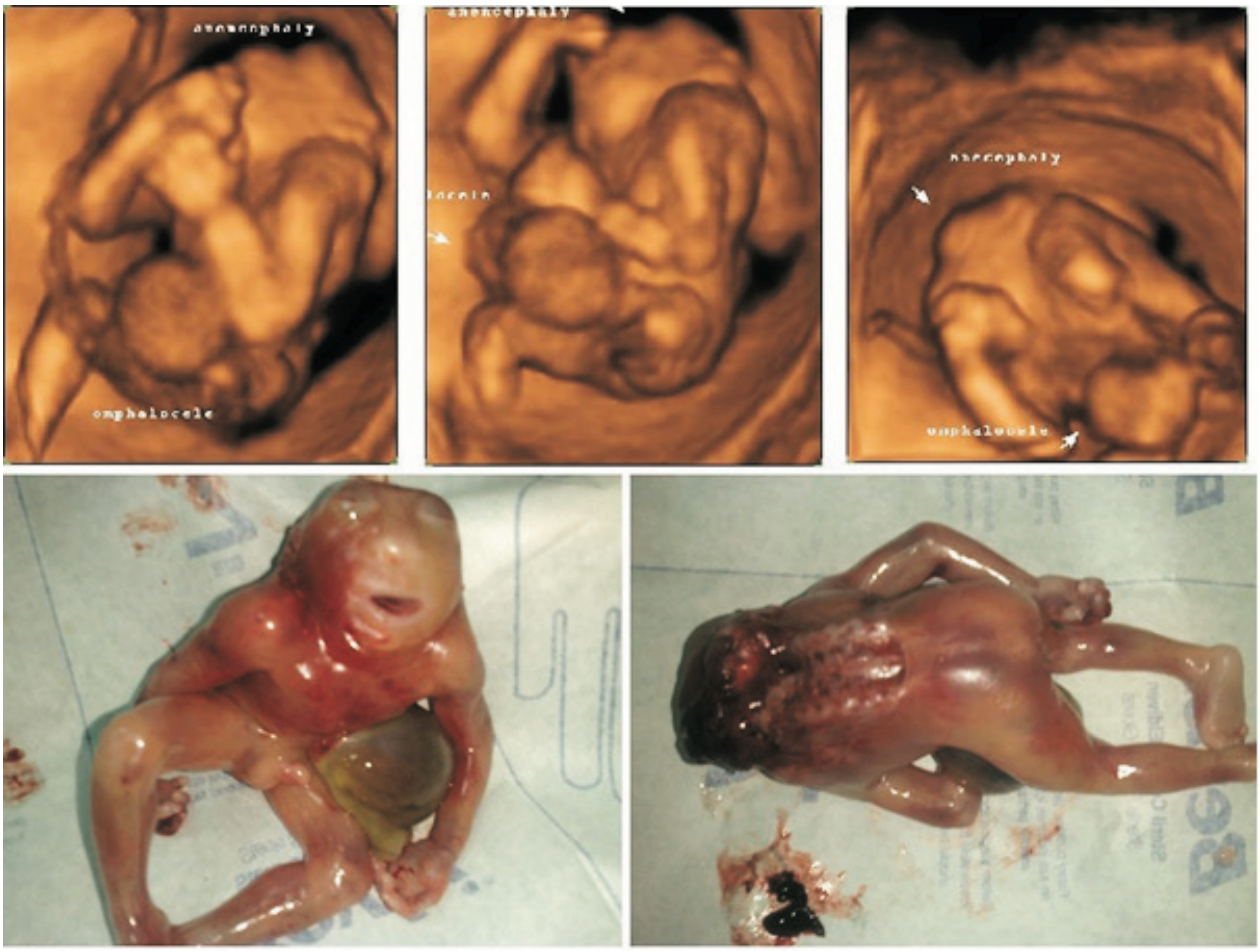

Figure 1.
Upper figures; 3D ultrasound images of the omphalocele and the anencephaly
Lower figures; the omphalocele and the anencephaly on the left side, and the anencephaly with the contiguous spinal defect (cranirachischisis totalis) on the right side.

Discussion

Central fusion failure at the umbilical ring, in babies with omphalocele due to defective mesodermal growth causes incomplete closure of the abdominal wall results in a persistent herniation of the midgut. The translucent sac, which is composed of amnion, Wharton jelly, and peritoneum, contains the abdominal viscera and the umbilical vessels radiate onto the sac wall. The extruded midgut is accompanied with the liver, spleen, and ovaries or testes in 50% of cases (1,3).

After fertilization around 18th days, the neural plate arises in the normal human embryo. The neural plate invaginates during the fourth week of development to form the neural groove. From the middle toward the ends in both directions, the neural tube is formed as closure of the neural groove progresses. Day 24 for the cranial end and day 26 for the caudal end are the completion days. Failures in normal closure process give rise to neural tube defects. Failure of neural tube closure at the cranial end of the developing embryo results anencephaly. Complete or partial absence of the brain and calvaria may be seen (5). A midline anterior abdominal wall defect, a herniated sac with visceral contents, and umbilical cord insertion at the apex of the sac are the prenatal US diagnostic features of omphalocele. Typically, the usual US appearance of anencephaly in the second and third trimesters is the absence of recognizable brain tissue. In the first trimester, anencephaly may appear relatively normal. However, absent cranium is the important US finding (6).

The association of omphalocele and central nervous system anomalies was observed in various situations. Most of them were genetical abnormalities. Large omphalocele with severe central nervous system anomalies was described in fetuses with trisomy (mostly trisomy 18) (6,7).

Associated karyotype abnormalities may also be diagnosed with the aid of ultrasound. Generally, the detection and the differential diagnosis rates of the two entities are high with conventional US, but isolated cases may be missed (2,8-10). Three-dimensional US may be helpful, especially in cases with difficulty to identify the exact pathology and to demonstrate the co-existing pathologies. In 3D ultrasound, the omphalocele is seen like a spherical mass protruding from the umbilical region of the fetal abdomen. The fetus looks like a baby holding a ball on the abdominal surface. In three-dimensional ultrasound, anencephaly is easily identified with the lack of brain and the skull above the supra-orbital line. In addition, three-dimensional ultrasound may help the family to understand and realize the situation (11). The thorough comprehension of the family will affect the decision process. The perception of the pathology, its prognostic value and the clinical management options can be made clear only after a realistic presentation of the anomaly. Following the exact perception of the situation the family can choose the options of the genetic counseling, pregnancy termination or continuation of the pregnancy (12).

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