CLINICAL CASE

Giant fetal omphalocele
Onfalocele fetal gigante

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ABSTRACT

Omphalocele, or exomphalos, is a congenital malformation of the midline of the abdomen involving herniation of abdominal contents at the base of the umbilical cord, limited by a membrane. It is rare and predominantly affects males. Early diagnosis of this pathology is currently possible, and ruling out other associated pathologies as well as markers of chromosomal abnormalities is essential for the prognosis of the fetus. We present a case of giant omphalocele with multidisciplinary management at the Belén de Trujillo Hospital, a tertiary care facility. Neonatology, pediatric surgery, and anesthesiology departments participated in the case, which was very important for a good outcome.

Keywords: Giant omphalocele, omphalocele, embryonic and fetal development.

RESUMEN

El onfalocele o Exónfalo, es una malformación congénita a nivel de la línea media abdominal con la herniación del contenido abdominal en la base del cordón umbilical, limitados por una membrana, poco frecuente con predominancia en el sexo masculino. El diagnóstico temprano de esta patología actualmente es posible, el descarte de otras patologías asociadas además de marcadores de cromosomopatía es fundamental en el pronóstico del feto. Presentamos un caso de onfaloce gigante co manejo multidisciplinario en un tercer nivel de atención en el Hospital Belén de Trujillo se contó con la participación en conjunto con neonatología, cirugía pediátrica y anestesiología y como en este caso fue muy importante para tener un buen desenlace.

Palabras clave: Onfalocele Gigante, Onfalocele, Desarrollo Embrionario y fetal.

Introducción

Omphalocele, or exomphalos, is a congenital malformation of the midline of the abdomen involving herniation of abdominal contents at the base of the umbilical cord, limited by a two-layered membrane (peritoneum and amnion)[1]. The incidence is 2.1–2.5/10,000 live births, with a slight predominance in males[2–4]. Omphaloceles can be classified: a) according to size: minor or major (small or giant) b) those that contain liver within the sac or not, and c) by location (epigastric, hypogastric, or umbilical)[5-7]. Isolated omphaloceles are rare, reported in only 14%[8] of cases, with 50-70% associated with other structural anomalies (most commonly cardiac and central nervous system anomalies) and chromosomal anomalies present in 30%[1]. Small omphaloceles without hepatic hernia are more commonly associated with chromosomal abnormalities[3]. The objective of this article is to present a very rare case of a giant omphalocele diagnosed prenatally (diameter of the opening greater than or equal to 5 cm)[5], with multidisciplinary management that resulted in closure of the defect and a favorable outcome. The importance of the case lies in the approach taken in the mother's womb, with genetic and structural ultrasound scans showing no structural abnormalities or markers for chromosomal abnormalities, leading the mother to make an informed decision to continue with the pregnancy and sign the informed consent form.

Case report

A 23-year-old primigravida patient with no known risk factors attends a private consultation for the first time. During a routine ultrasound scan, a protrusion is observed at the insertion of the umbilical cord, and she is referred to a tertiary hospital for a genetic ultrasound scan, which reports: Crown-rump length (CRL): 82 mm, nuchal translucency of 1 mm, nasal bone present, no tricuspid regurgitation observed, venous ducts cannot be performed due to the mass protruding at the cord insertion covered by a membrane, with presence of the liver, no other structural alterations observed, calculating the risk for chromosomal abnormality to be less than 1/1000. The patient attends two prenatal check-ups, at 23 weeks and 33 weeks of gestational age, where ultrasound reveals and confirms the diagnosis of a defect at the base of the umbilical cord insertion measuring 10 cm (see Figure 1), with no other structural abnormalities present.

At 34 weeks of gestational age, she was admitted as an emergency case due to uterine dynamics, which subsided after 48 hours of administering corticosteroids for fetal maturation. The case was referred to neonatology and pediatric surgery. Uterine dynamics persisted and she went into labor, culminating in a cesarean section, resulting in a male infant weighing 2000 g, Apgar scores of 9 and 10, an umbilical hernia containing small intestine and liver with a 7 cm opening at the base (see Figure 2), He was admitted to the neonatal intensive care unit, intubated and sedated with fentanyl, a muscle relaxant (vencuronium) was used, and the defect was covered with a hydrogel silo (see Figure 3), with the contents being introduced progressively each day.

Figure 1. Ultrasound shows a defect at the base of the umbilical cord insertion.

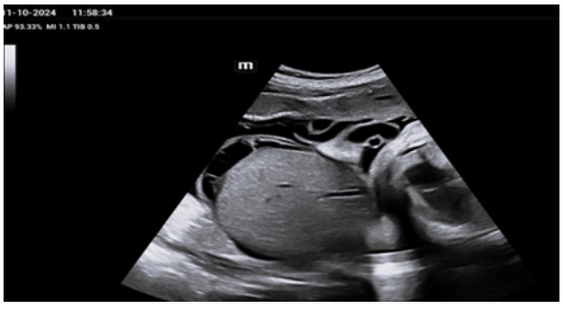


Figure 2. Umbilical hernia with small intestine and liver contents, with a 7 cm opening at the base.

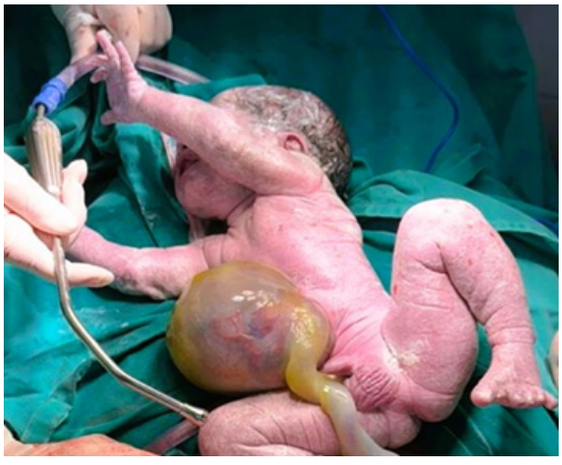
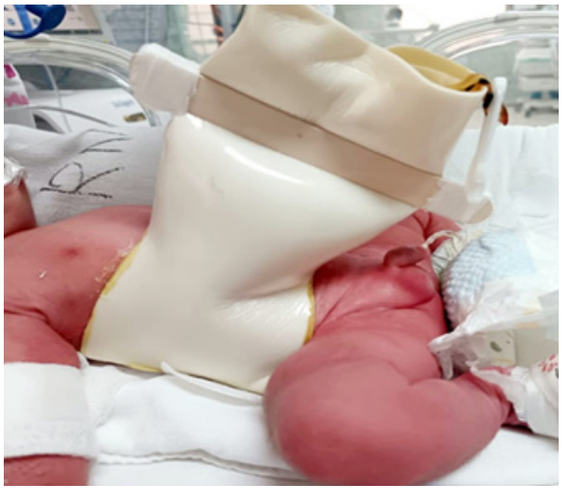


Figure 3. The defect is covered with a hydrogel silo.



Progress

Seven days after birth, he underwent surgery to close the defect in stages, followed by sedation and relaxation for seven days. The relaxation and sedation were then gradually withdrawn over the 14 days following the surgery (see Figure 4).

After that, she was discharged, breastfeeding. She was monitored in the following months, reporting no discomfort or incidents.

Discussion

Many strategies have been described for giant omphaloceles, but there is no standard of care.

The management of giant omphalocele can be summarized as surgical and non-surgical closures. Staged surgical closure techniques include different types of sutured silos, intra-abdominal

Figure 4. Evolution. Seven days after birth, the defect is closed in stages, continuing with sedation and relaxation.

tissue expanders, and closure techniques such as synthetic mesh interposition, skin flaps, skin grafts, and dermal matrix. All of these involve multiple interventions under anesthesia. The ultimate goal is to close the abdominal wall fascia or cover the liver and intestine with skin. In this case, as described in the literature, the Duoderm silo was used[7].

In our case, the defect is classified as a large or giant omphalocele, or omphalocele with extracorporeal liver, based on its size, location, and content. Two-dimensional (2D) ultrasound has a specificity greater than 97% and a sensitivity of 60-80%, the latter being affected by the position of the fetus, the presence of other defects, and the experience of the operator[9]. In our case, no association was found with other malformations or other pathologies reported as transposition of the great vessels, pulmonary hypertension, or congenital heart disease, as described in the literature in 37.5% of cases[7]. The etiology of omphalocele, as with most congenital anomalies, is unknown.

Factors such as exposure to teratogenic agents such as serotonin receptors in the first weeks of gestation, nutritional deficiencies, and genetic predisposition possibly linked to maternal enzyme defects, maternal age over 30 years, and black race are postulated. There were no risk factors, and it is consistent with the literature that cases are sporadic with a predominance of males[3,11]. Early prenatal diagnosis and timely referral to a tertiary care center, where a joint decision on care is made, is associated with a good prognosis[12,13].

The survival rate for isolated omphalocele is 90%. Even so, monthly ultrasound monitoring is recommended. There is controversy regarding the mode of delivery. The absence of randomized studies makes it difficult to clearly recommend one mode of delivery over another. Despite the controversy, most authors advocate elective cesarean section in cases of abdominal defects greater than 5 cm and with hepatic content, due to the risk of abdominal visceral trauma and/or rupture of the hernial sac. In our case, the method of choice was cesarean section. The staged non-surgical management of giant omphaloceles with silos is safe, easy, and economical. It shortens the closure time and reduces the potential morbidity and mortality compared to traditional surgical or medical management. For the reasons described above, interdisciplinary management is essential for the prognosis of these cases.

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