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ESTUDI DE CASOS

Cloacal exstrophy, a rare fetal malformation with difficult prenatal sonographic diagnosis: two case reports

Extrofia cloacal, una malformación fetal rara con diagnóstico ecográfico prenatal difícil: dos informes de casos

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Abstract

Cloacal exstrophy is a rare condition for which prenatal diagnosis is challenging. This pathology entails a great deal of fetal and neonatal morbimortality that requires multidisciplinary management.

We present two cases of perinatal diagnosis. The first of these is a case in which the cloacal exstrophy was diagnosed prior to the rupture of the cloacal membrane, which is rare in practice and hardly described in the literature. In the second case, an abnormality of the non-filmed abdominal wall was detected, which had a fatal outcome, the death of the newborn. The diagnosis came after, by autopsy study of the cloacal exstrophy.

Keywords: Fetal malformations, prenatal diagnosis, antenatal care, obstetrics sonography, cloacal exstrophy.

Resumen

La extrofia cloacal es una condición poco común para la cual el diagnóstico prenatal es un desafío. Esta patología conlleva una gran cantidad de enfermedades fetales y morbimortalidad neonatal que requiere un manejo multidisciplinario.

Presentamos dos casos de diagnóstico perinatal. El primero de ellos es un caso en el que la extrofia cloacal fue diagnosticada antes de la rotura de la membrana cloacal, que es rara en la práctica y poco descrita en la literatura. En el segundo caso, se detectó una anomalía de la pared abdominal no filmada que tuvo un desenlace fatal, la muerte del recién nacido. El diagnóstico vino después por estudio de autopsia de la extrofia cloacal.

Palabras clave: malformación fetal, diagnóstico prenatal, atención prenatal, ecografía obstétrica, extrofia cloacal.

Introduction

Cloacal exstrophy is defined as a complex anomaly that affects the urogenital and intestinal tracts. It is the most serious form of anomalies that are described within the so-called exstrophy-epispadias complex[1].

Its prevalence is 2-4/100,000 births[1-3]; although it is believed that this value may be underestimated, being the real incidence 1 per 10,000-50,000 pregnancies[3].

Its prognosis varies depending on the anomalies associated[1]. Historically, it was considered a devastating pathology due to complications such as sepsis, hydroelectrolytic alterations and nutritional deficits associated with short bowel syndrome or intestinal obstructions. However, currently, progress in neonatal care and surgical techniques have allowed achieving survival rates of between 83 and 100%[3].

Case presentations

Patient 1

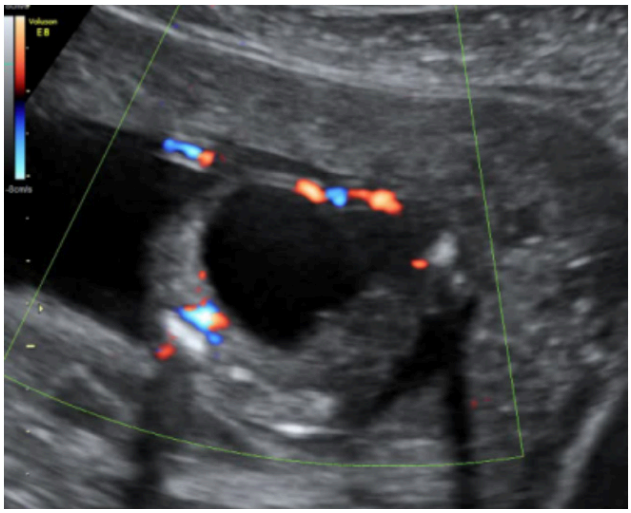
A 28-year-old Spanish woman, who was gravid 1, para 0, was referred for obstetrics ultrasound scan for dating and for the first trimester screening in her 12th week of pregnancy. There was nothing relevant in her medical history and she was not receiving medication during this pregnancy.

Transabdominal ultrasound examination showed a singleton live intrauterine foetus with an abnormal abdominal anechogenic image. For that reason, we scheduled an appointment two weeks after to check the evolution and the screening results which showed low risk of chromosomopathies.

In the following ultrasound, the foetus showed an abdominal econegative image, which was surrounded by the

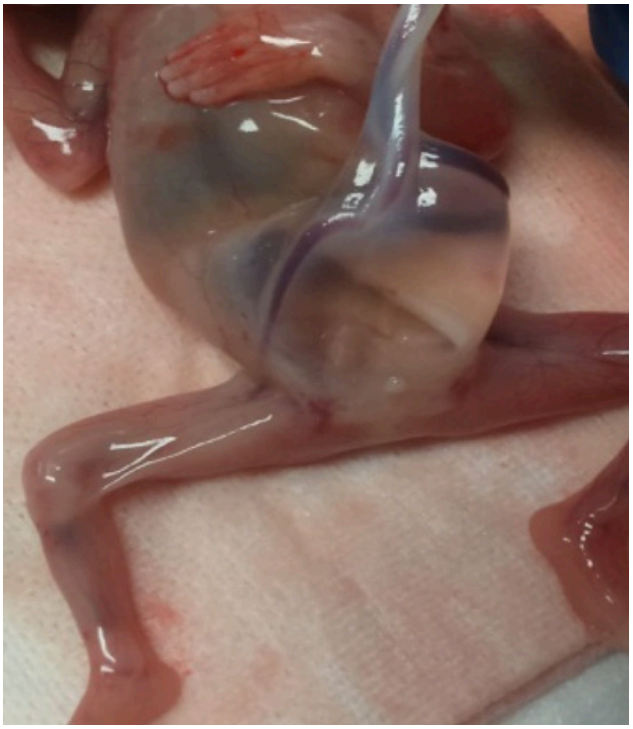
umbilical arteries, without visualization of the rectovesical interface (**Figure 1**). In addition, a hyperechogenic left kidney was evident without the possibility of visualizing the right kidney. Male genitalia were observed.

Figure 1: Ultrasound in week 15 of gestation where the large suspicious econegative image of cloacal exstrophy is observed.



The sonographic suspicion was of a cloacal exstrophy, multidisciplinary approach was performed to advise the patient and she decided to terminate the pregnancy. When she delivered the fetus the defect of the anterior wall was observed (**Figure 2**). The fetus and placenta were referred for anatomopathological study and the diagnosis was confirmed.

Figure 2: The fetus images once he was delivered. We can observe the defect in the anterior abdominal wall.



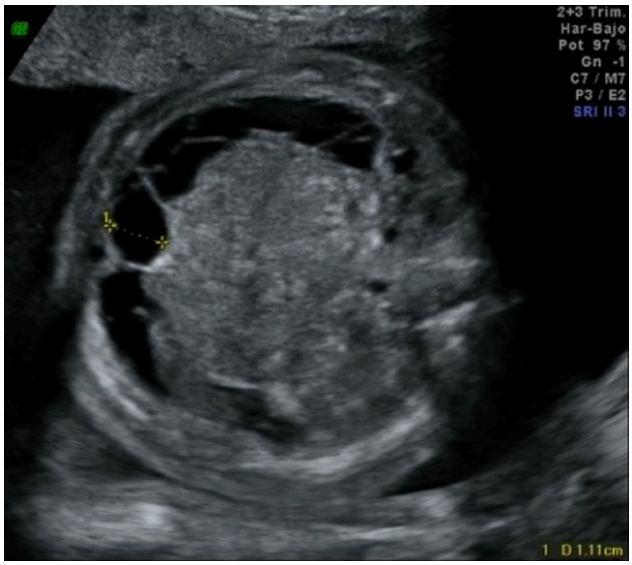
Patient 2

Our second patient was a 38-year-old, nulliparous, Caucasian woman with a non-consanguineous Caucasian partner, no significant medical or family precedent and stated no illicit substance use. She appeared with an uncomplicated pregnancy with a low-risk screening result on nuchal translucency for aneuploidy. At the 20th-week foetal anomaly morphology scan, a unique umbilical artery and ascites were identified as highlights. It was accordingly decided to perform an amniocentesis, which resulted in a 46XX karyotype. Serologies, arrays of metabolic diseases, as well as studies for cystic fibrosis were negative.

In subsequent ultrasound controls, a moderate progression of ascites was observed, which showed a tabulated appearance, without other accompanying findings. An abdominal cystic lymphangioma was proposed as the first diagnostic alternative.

In week 33 + 3 she was referred to our centre after a premature rupture of membranes. Before its diagnostic confirmation, the pattern of fetal lung maturation with betamethasone began. Ultrasound showed a multicabicated intra-abdominal cystic image that occupied the entire cavity (**Figure 3**).

Figure 3: 30 weeks sonography demonstrated ascites with presentation of partitions with a maximum thickness of 11mm.

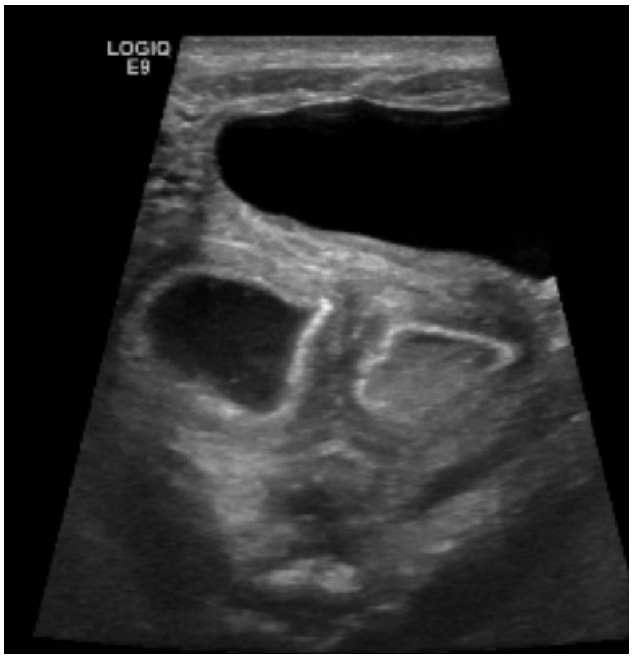


After evidencing deep spontaneous decelerations and a decrease in the variability in the cardiotocographic registry, an urgent caesarean section was indicated, delivering a live girl, Apgar 6/8 that required an admission into Neonatal Intensive Care Unit.

The baby weighed 1865 g and showed a large abdominal distension with respiratory compromise, which required orotracheal intubation. Large cystic mass in lower

hemiabdomen, ambiguous genitals, absence of anus with a single hole, and anal fosse in anterior situation. Uncrossed interglute groove. A common channel was channelled with an abundance of urine and detritus.
Postnatal ultrasounds showed a meconial pseudocyst without dilation of the rectum, suggesting cloaca and / or fistula. Sacrococcygeal and medullar dysgenesis and uterine and vaginal duplicity (**Figure 4**). Bilateral pyeloureteral dilation. The colon biopsy showed presence of ganglion cells.

Figure 4: Postnatal sonography demonstrating an uterine and vaginal duplicity.



On the second day of life, a laparotomy was performed due to the suspicion of a cloacal malformation with fibroadhesive meconial peritonitis. During surgery, there was evidence of intestinal perforation and massive hydrosalpingometrocolpos that was decompressed by colpostomy (possibly meconial peritonitis was due to meconium refluxing the genitourinary system). The progressive instability of the patient forced closure (no tension with colostomy).

At 10 days of life, the patient was reoperated due to intestinal perforation, with resection of the necrotic jejunum and terminal-terminal anastomosis. The patient suffered progressive deterioration, with deterioration of the hemodynamic state, pulmonary emphysema and sepsis. At 21 days of age, a limitation of therapeutic effort was agreed with relatives what led to Exitus.

The necropsy confirmed the existence of a severe persistent anus-rectal malformation with agenesis / atresia of the anus-rectum, double uterus and vagina, sacral agenesis, fusion defect / partial absence of L4, inferior spinal dysgenesis, in addition to hypertrophy and right ventricular dilation with patent foramen ovale, tricuspid valve dilation and signs of acute right heart failure with edema and pulmonary haemorrhage, congestive hepatosplenomegaly and cholestatic hepatitis with jaundice.

Discussion

Cloacal exstrophy is a rare congenital anomaly that occurs in a 0.25-0.5 / 10,000 births[1,2] being very few cases diagnosed prenatally.

The pathology at hand is the most serious form of a spectrum of anomalies known as the exstrophy-epispadias complex, which includes different malformations with varying degrees of severity, ranging from epispadia and bladder exstrophy, to cloacal exstrophy[2]. Bladder exstrophy is the most common form, occurring in 0.3 / 10,000 births, followed by epispadia, which occurs in 0.08-0.25 / 10,000[1].

Its aetiology is currently unknown, although it is believed that it could be due to unknown genetic and environmental factors[4]. A ratio of 2:1 for males has been described[3].

Embryology

In order to understand the spectrum of the exstrophy-epispadias complex, it is essential to know part of embryology.

The cloacal membrane is a transient structure that is consisting of endoderm and ectoderm and is located anterior to the cloacal bag. Around week 4-5 of gestation, two mesoderm migrations occur, in a caudal direction to the navel that will form the infraumbilical wall and genital tubercle and the other migration that will be placed in the urogenital septum that divides the cloacal bag into the urogenital sinus and rectum in week 7-8 of gestation, with the subsequent rupture of the cloacal membrane around week 8 of gestation[3-6].

An anomalous development of this process will lead to persistence and subsequent rupture of the cloacal membrane and will, depending on the moment in which it occurs, give rise to the pathologies described in the exstrophy-epispadia complex. In cloacal exstrophy, a rupture of the cloacal membrane has been described prior to the fusion of the urogenital septum. However, there are several clinical cases in the literature, since 1992, which describe an abnormal persistence of the cloacal membrane in the context of exstrophy until week 20-22sg with subsequent rupture of the same and diagnosis of cloacal exstrophy[1,7,8,9,10]. In our first case, we also suspected cloacal exstrophy prior to rupture of the cloacal membrane in week 15 (due to the visualization of an anechogenic, cystic image on the abdominal wall), and the omphalocele was observed in the post-mortem analysis.

Prenatal diagnosis

For its prenatal diagnosis there is the ultrasound tool. Its diagnosis is complex and therefore it is rare. The data of a study based on a series of specific cases showed that only 25% of the effects can be accurately diagnosed in the prenatal period[6,11]. MRI and karyotype can provide additional useful information[12,13].

Cloacal exstrophy is the anomaly of the exstrophy-epispadias complex that usually carries the most associated malformations. This abdominal anterior wall defect is usually accompanied, in 70-90% of cases, by an omphalocele of variable size. The association of omphalocele, cloacal exstrophy, imperforate anus and spinal anomalies is known as the OEIS complex[1,6,14].

It has been described that 100% of patients with cloacal exstrophy suffer from an alteration resulting from a defective closure of the neural tube[4]. We can also see abnormalities at urological level, the most frequent described are the ectopic pelvic kidney, the ureterocele and the horseshoe kidney (5) and the hemibladder and abnormalities at the digestive level[3]. In the genitals we can also observe alteration from the type of uterine and / or vaginal duplication in women, as well as bilateral cryptorchidism in men[4].

As described above, there are cases in which the anomaly is detected prior to the rupture of the cloacal membrane. In these cases, we would see the non-visualization of the bladder, a defect of the infraumbilical anterior wall of the large midline or the structure cystic anterior wall (persistent cloacal membrane), omphalocele and lumbosacral abnormalities[1,7].

Management

After an early diagnosis of cloacal exstrophy, it is necessary to carry out a multidisciplinary management that must be initiated in the prenatal period, for an adequate advice to parents.

When clinical suspicion is prior to fetal viability, legal termination of pregnancy should be offered[1]. If the parent swish to continue with the pregnancy, they should be informed that the affected newborn will require immediate multidisciplinary postnatal care, as well as the beginning of a cascade of surgical interventions that will usually begin in the early postnatal period, with an immediate closure of the meningocele and omphalocele if present, as well as the two hemibladders, so they should delivered in a hospital with neonatal intensive cares and facilities for paediatric surgery. In a second phase, the bladder and bowel will be reconstructed in consecutive phases[6,15].

The improvement in neonatal care, as well as instruments and surgical techniques have led to a drastic increase in survival rates, as well as improvement in continence results and, therefore, in the quality of life[15]. Currently, survival rates are close to 100% and the most important principles of its management include adequate nutritional support, early closure of the exstrophy and preservation of intestinal length. The achievement of urinary and faecal continence remains a challenge[3]. The Fetal Medicine Foundation describes that both, males and females, with this condition are capable of normal lifestyle and both sexes are fertile after surgery. Some form of urinary tract diversion is required for all.

Even so, patients with such a diagnosis will require long-term follow-up by a multidisciplinary team, which will also include psychosocial aspects[6], because the anomalies involved with this alteration require major surgical reconstructions with implications in continence, renal function, appearance and genital function.

Disclosure of interests

Full disclosure of interests available to view online as supporting information.

Contribution to authorship

All individuals that qualified for authorship have been included and all those included qualify for authorship. Miriam Crespo and Laia Vila Homs contributed to conception and design, interpreted the data and drafted the article. Rafael José helped with the draft. Jose Luis Vidal and Rosa Gopegui were the reviewers of the article and Celia Garrido and Carmen Simón where the Consultants in charge. All authors revised the article critically for intellectual content and approved the final version to be published.

Details of ethics approval

The patient gave verbal and written informed consent for this report, which conforms to the ethical guidelines of the Helsinki Declaration of 1975.

Details of ethics approval

None.

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