Prenatally diagnosed segmental intestinal dilatation associated with anorectal malformation

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ABSTRACT

Introduction. Congenital segmental intestinal dilatation has a low incidence within the differential diagnoses of fetal abdominal cyst masses. Suspicion may arise at prenatal ultrasonography, but diagnosis is confirmed at surgery and subsequently at histopathological analysis. There are various theories available to explain its etiopathogenesis. Association with anorectal malformations is rare.

Clinical case. Newborn prenatally diagnosed with an abdominal cystic mass and diagnosed at birth with an associated anorectal malformation, with postoperative confirmation of segmental intestinal dilatation.

Discussion. Segmental intestinal dilatation should be considered within the differential diagnoses when an abdominal cystic mass is prenatally detected at ultrasonography. Association with anorectal malformations at birth has been described, but it is unusual. Suspicion allows diagnosis and adequate treatment to be established.

KEY WORDS: Digestive system abnormalities; Anorectal malformations; Ultrasonography, prenatal.

DILATACIÓN SEGMENTARIA DE INTESTINO DELGADO DE DIAGNÓSTICO PRENATAL ASOCIADA A MALFORMACIÓN ANORRECTAL

RESUMEN

Introducción. La dilatación segmentaria intestinal congénita es una patología de baja incidencia dentro de los diagnósticos diferenciales de masas quísticas abdominales fetales. Su sospecha inicial puede surgir en la ecografía prenatal, aunque su confirmación diagnóstica se realiza durante la cirugía y posteriormente en el análisis histopatológico. Existen diversas teorías acerca de su etiopatogenia. Su asociación con las malformaciones anorrectales es de baja frecuencia.

Caso clínico. Paciente neonato con diagnóstico prenatal de masa quística abdominal con diagnóstico al nacimiento de malformación anorrectal asociada y la confirmación postquirúrgica de una dilatación segmentaria intestinal.

Comentarios. La dilatación segmentaria intestinal debe ser considerada entre los diagnósticos diferenciales ante el hallazgo ecográfico prenatal de una masa quística abdominal. Al nacimiento, su asociación con malformaciones anorrectales está descrita aunque es inusual. Su sospecha permite el diagnóstico y su adecuado tratamiento.

PALABRAS CLAVE: Malformación del sistema digestivo; Malformación anorrectal; Ecografía prenatal.

INTRODUCTION

Intra-abdominal cystic masses are found at prenatal ultrasonographies in the second or third pregnancy trimesters. Segmental intestinal dilatation (SID), which is unusual, is one of them[(1)]. It can be macroscopically confirmed at surgery and microscopically confirmed at histopathological analysis[(2)]. Various theories regarding etiology and association with other pathologies have been proposed. Association with anorectal malformations (ARMs) has been described, but it is rare[(3)].

We present the case of a newborn prenatally diagnosed with an intra-abdominal cystic mass and diagnosed at birth with an associated ARM, with postoperative confirmation of SID.

CLINICAL CASE

Male patient, pregnancy under control. At routine ultrasonography in the third pregnancy trimester (week 29), a 17.4×12×22.3 mm anechoic image with regular borders and thin echoes within was observed (Fig. 1). In the physical exploration at birth, the abdomen was soft and non-tender, with no evidence of palpable masses or color changes. At the perineal level, an anorectal malformation

DOI: 10.54847/cp.2024.04.16

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Date of submission: March 2023 *Date of acceptance:* September 2024

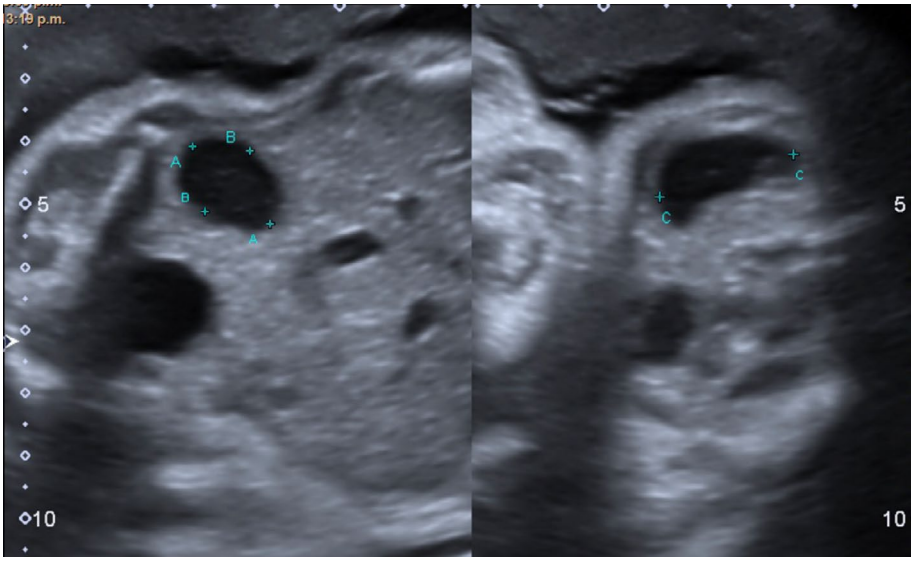

Figure 1. Ultrasonography at prenatal week 29: 17.4 × 12 × 22.3 mm diameter anechoic image with regular borders and thin echoes within.

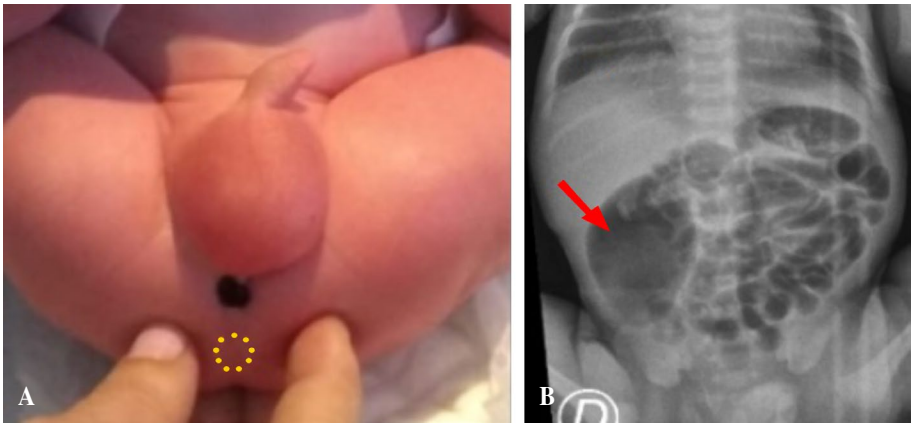

Figure 2. A) Anorectal malformation with perineal fistula. Spontaneous meconium release is observed (the *dotted circle* marks the site of the theoretical anus; the sit bones are marked with the fingers). B) Abdominal X-ray: aerated cystic mass *(arrow)*.

with a perineal fistula in the scrotal base was noted, with little meconium release at that level (Fig. 2A).

Associated pathologies were searched for by means of metabolic studies, an echocardiogram, a brain and a kidney ultrasonography, a vertebral and a sacral X-ray, an ocular fundus examination, and a karyotype study (XY), all of them with normal results. Abdominal X-ray revealed a radiolucent image in the right hemiabdomen (Fig. 2B).

A study of the colon through water-soluble contrast enema was requested. The colon was normal in caliber, but it was displaced to the left by a space-occupying structure (Fig. 3).

In light of the progressive abdominal distension present, with no effective stools, surgery was decided upon on the second day of life. An exploration laparoscopy was carried out. Ileal dilatation 30 cm away from the ileocecal valve was observed. The latter was exteriorized through the umbilical port. It was 8 cm in length, and it had a caliber 10 times greater than a normal ileum (Fig. 4). Segmental resection was eventually decided upon, and an end-to-end anastomosis and a complementary appendectomy were carried out. The ARM was subsequently repaired by means of a minimal posterior sagittal anorectoplasty.

The pathological report demonstrated a submucosal edema and a thickened muscularis propria, suggestive of ischemia, with presence of lymph node cells.

The patient had digestive transit 48 hours later, and enteral nutrition was initiated on postoperative day 5. The patient was discharged 15 days later. Outpatient controls demonstrated adequate enteral tolerance and weight and size progression. Today, sphincter control is normal.

DISCUSSION

When a cystic mass is found prenatally, accurate ultrasound diagnosis may prove challenging, since the mass may be indicative of various pathologies. In spite of its low incidence, SID should be considered within the differential diagnoses and monitored until birth[4]. In this patient,

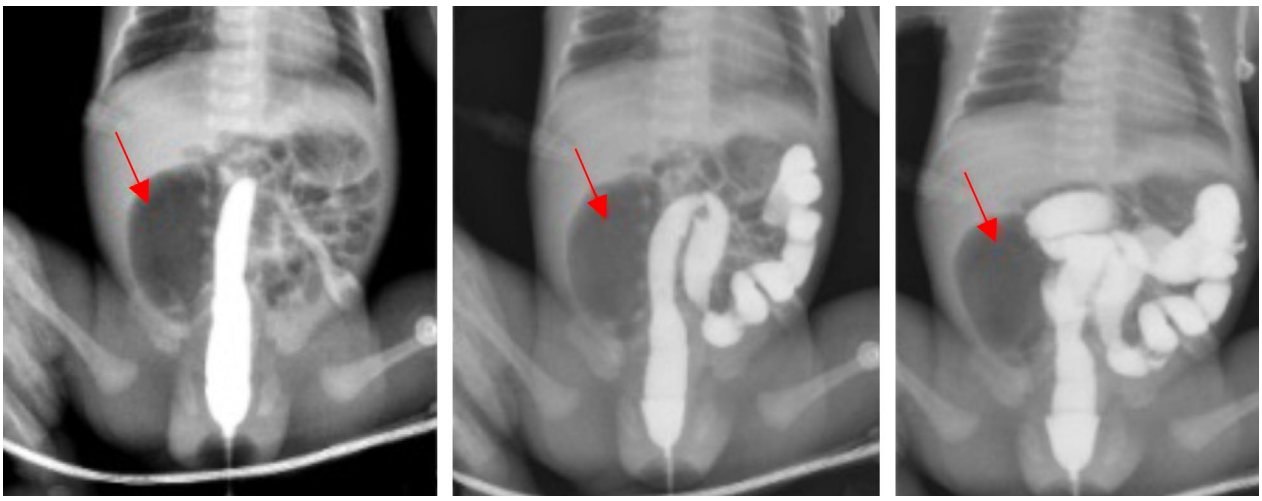

Figure 3. Water-soluble contrast enema. The association with intestinal dilatation is to be considered *(arrow)*.

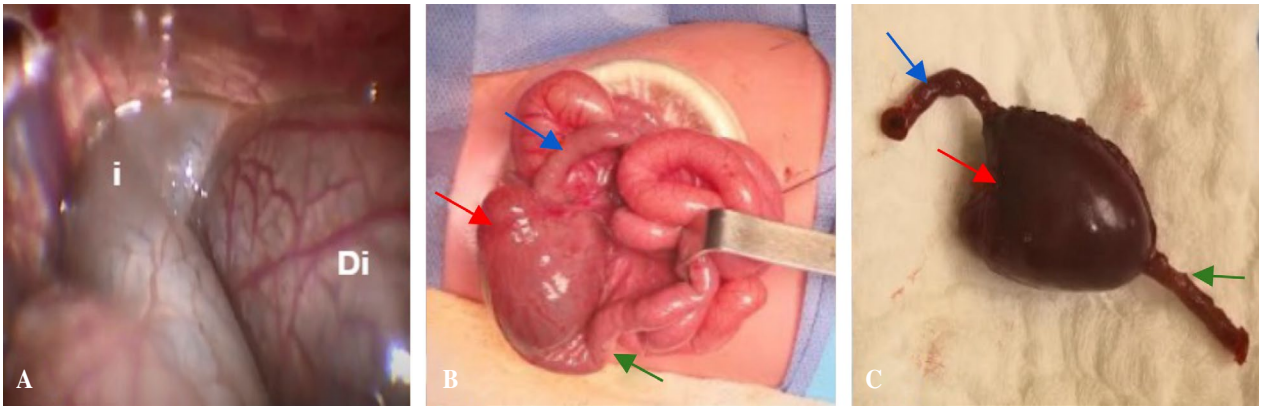

Figure 4. Intraoperative images. A) Exploration laparoscopy (*i:* normal small bowel; *Di:* dilated small bowel). B) Umbilical exteriorization. C) Intestinal specimen submitted to the pathology department. *Red arrows:* intestinal dilatation; *green arrows:* proximal small bowel; *blue arrows:* distal small bowel.

a cystic lesion was found at ultrasonography in the third pregnancy trimester, with suspected intestinal dilatation or duplication cyst, given its location.

SID was first described by Swenson and Rathauser in 1959, with accurate and identifiable diagnostic criteria at surgery and subsequent histopathological analysis. These criteria include dilatation limited to an intestinal sector, with intestinal size being 3 or 4 times greater than usual, and with an abrupt transition between the normal and the dilated parts. There should not be an intrinsic or extrinsic occlusion distal to the dilatation, even if the patient has clinical signs of occlusion/subocclusion. Intestinal nervous plexuses should be normal, with complete functional recovery following the resection of the segment involved. It is defined as "definitive" when all criteria are met, and as "potential" if only anatomical criteria are[5].

In our patient, surgical exploration revealed what could correspond with a localized SID at the level of the distal ileum, which means the aforementioned anatomical diagnostic methods were met.

In a meta-analysis of 150 patients with segmental intestinal dilatation from 1959 to 2020, the most frequent locations of the lesion were the ileum (56%) and the colon (27.3%), followed by the jejunum (8.7%) and the duodenum (6.7%). 57.3% of the patients had associated abnormalities, most frequently involving the digestive system (69.8%), the abdominal wall (19.8%), and the cardiovascular system (11.6%). In terms of the digestive system, intestinal malrotation was the most frequent, followed by anorectal malformation, which was less common[6].

Our patient had a normal vertebral X-ray and a normal echocardiogram at birth, with the postnatal finding of ARM associated with the image of prenatal cystic dilatation.

The etiopathogenesis of SID remains somewhat unclear today. The main hypotheses include intestinal strangulation at the umbilical ring in the fetal period, tortuous vessels

extrinsically compressing the intestinal wall, prenatal intestinal ischemia leading to hypoplastic smooth muscles, "kinking" or mesenteric volvuli, and even interruption of the intestinal nervous plexus in this area, which differs from zonal aganglionosis (Hirschsprung-like disease) in that the nervous plexuses should be normal for the pathology to be considered a SID[7].

Our case can be somewhat explained by the theories currently in force regarding the origin of the malformations found (SID, ARM), in order to understand embryological relationship. The injuries could have stemmed from associated systemic vascular compromise as a result of inadequate organogenesis. Specifically, intrauterine obliteration of the ileocolic branch of the superior mesenteric artery could have led to segmental dilatation, whereas intrauterine obliteration of the inferior mesenteric artery could have led to ARM[3].

When reviewing this case, the authors found certain diagnostic and treatment aspects that were regarded as opportunities for change and improvement. First, the enema was conducted with water-soluble iodinated contrast, without assessing retrograde peristalsis and verifying whether defect contrast was positive or not. Using barium as a contrast would have allowed us to gather further information in this respect. In addition, there were no profile images available, which would have allowed the ARM to be assessed in a more accurate fashion. Second, resecting the cecal appendix during surgery might not have been a good idea, since an ARM patient like this could have benefitted from having it in case of poor progression regarding fecal or urinary continence.

In conclusion, when a cystic mass is found at prenatal ultrasonography, segmental intestinal dilatation should be considered within the differential diagnoses. At birth, association with anorectal malformations is rare, but does occur. In our case, the fact it was found prenatally and monitored until birth, along with early treatment, allowed definitive diagnosis to be achieved, thus providing the patient with an effective treatment.

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