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Intestinal malrotation is a rare congenital condition with an incidence in adulthood between 0,0001% and 0,19%, affecting nearly 1:500 live births. It results from an abnormal rotation of the bowel within the peritoneal cavity during embryogenesis. Generally it involves both small and large bowel, leading to an increased risk of intestinal obstruction.
Depending on which phase of midgut embryological development is stopped or disrupted, a variety of anatomic anomalies may occur. Reverse rotation is the most rare form of intestinal malrotation (2-4%) and is more common in women. It origins from premature return of the caudad midgut into the abdominal cavity while the duodenal loop rotates clockwise during fetal life, between 4th and 12th gestational week. The cecum begins its migration and shifts to the right behind the superior mesenteric artery (SMA). As a result the transverse colon lies behind the duodenum and the SMA. Malrotation's most common clinical manifestations in neonates are acute duodenal obstruction and midgut volvulus, life-threatening conditions resulting in acute bowel obstruction and ischemia. In adult patients the risk of volvulus decreases and clinical presentation is more aspecific, leading to delayed diagnosis, that may cause dangerous consequences.
We report a rare case of an adult male patient presenting with acute abdominal symptoms caused by a reverse midgut rotation in a Beckwith-Weidemann Syndrome (BWS), a rare genetic disorder characterized by the association between adrenal cytomegaly, hemihypertrophy, macroglossia, omphalocele and pancreatic islet hyperplasia.

KEY WORDS: Beckwith-Wiedemann syndrome, Reverse midgut rotation, Jejunal transmesenteric hernia

Introduction

Intestinal malrotation is a rare congenital condition with an incidence in adulthood between 0,0001% and 0,19%, affecting nearly 1:500 live births [1,2]. It results from an abnormal rotation of the bowel within the peritoneal cavity during embryogenesis. Generally it involves both small and large bowel, leading to an increased risk of intestinal obstruction [3].

Depending on which phase of midgut embryological development is stopped or disrupted, a variety of anatomic anomalies may occur. Reverse rotation is the most rare form of intestinal malrotation (2-4%) and is more common in women. It origins from premature return of the caudad midgut into the abdominal cavity while the duodenal loop rotates clockwise during fetal life, between 4th and 12th gestational week. The cecum begins its migration and shifts to the right behind the superior mesenteric artery (SMA). As a result the transverse colon lies behind the duodenum and the SMA[13]. Malrotation's most common clinical manifestations in neonates are acute duodenal obstruction and midgut volvulus, life-threatening conditions resulting in acute bowel obstruction and ischemia. In adult patients the risk of volvulus decreases and clinical presentation is

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more aspecific, leading to delayed diagnosis, that may cause dangerous consequences [4,13].
We report a rare case of an adult male patient presenting with acute abdominal symptoms caused by a reverse midgut rotation in a Beckwith-Weidemann Syndrome (BWS), a rare genetic disorder characterized by the association between adrenal cytomegaly, hemihypertrophy, macroglossia, omphalocele and pancreatic islet hyperplasia [5].

Case Report

A 28 y/o patient was admitted to our Emergency Department for severe central abdominal pain associated to nausea and absence of flatus from 24 hours.
Patient history revealed an uncommon story of BWS: the patient underwent a glossoplasty for macroglossia, and a hernioplasty for umbilical hernia during neonatal life, and MRI follow-up for multiple renal cysts from 2013. He reported that in the last years he suffered from recurrent abdominal pain associated with diarrhea and meteorism. A colonoscopy had been performed for his symptoms with no abnormal findings.
At clinical examination the patient was hemodynamically stable and afebrile. The abdomen was diffusely tender and painful in the central quadrant, no signs of rebound.

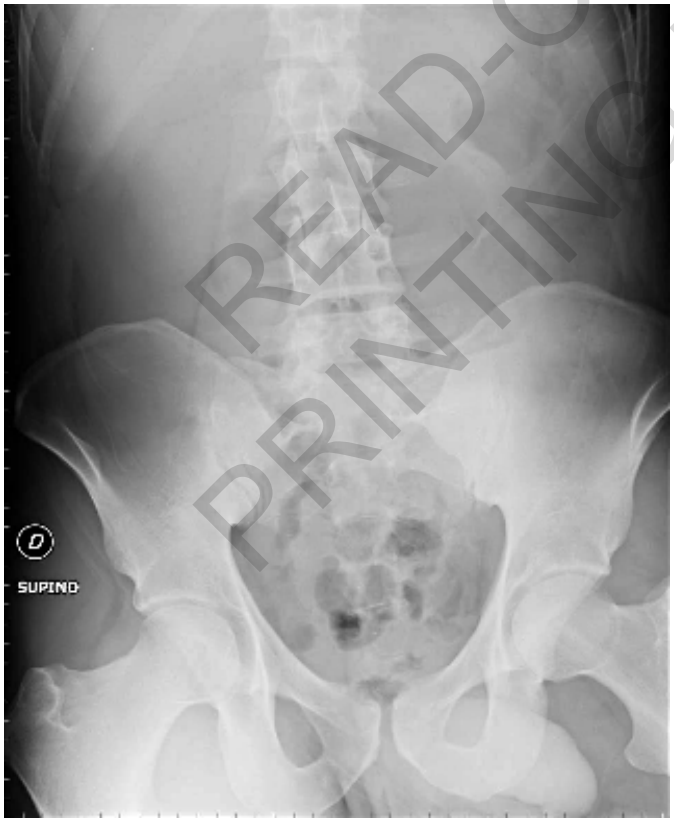

Fig. 1

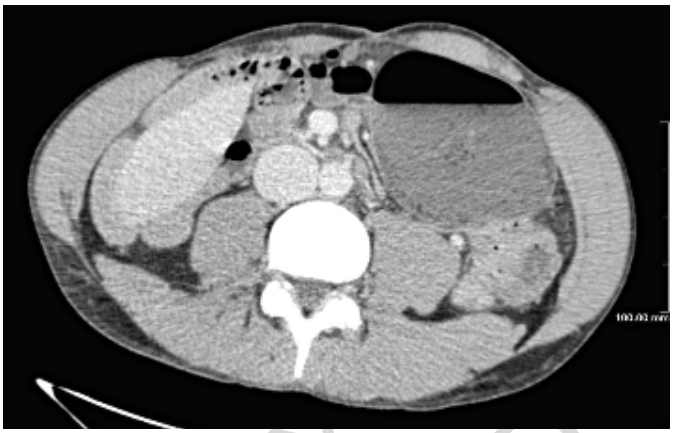

Fig. 2

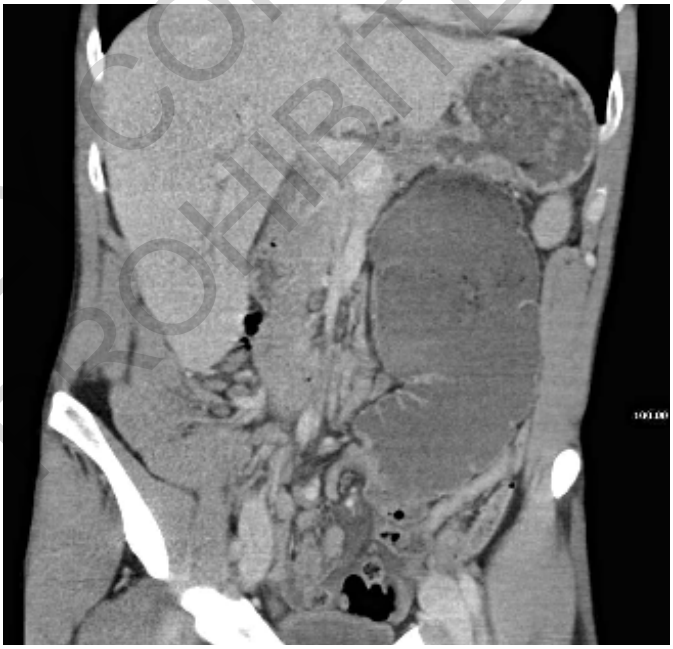

Fig. 3

Blood tests revealed only a modest increase of CRP (16,5 mg/L). The patient was managed with resuscitation fluids and analgesic therapy. In the meantime abdominal X-rays showed no signs of perforation but a small and large bowel dilation with absence of colonic profile in the right abdominal quadrants (Fig. 1). An abdominal CT-scan was performed: it showed abnormal bowel dilation (10 cm left-sided cecum) with abrupt intestinal obstruction (bird's beak sign), a medialized duodenum, and inversion of SMA and superior mesenteric vein (Figs. 2, 3).
Due to incoming hemodynamic instability the patient required emergency surgery, in the suspect of bowel occlusion in intestinal malrotation. A laparotomy was performed: the cecum appeared dilated (10 cm diameter) and positioned in the upper left quadrant of the

abdomen. The duodenum was placed more medial than usual, and there wasn't any Treitz ligament, confirming the diagnosis of reverse midgut rotation. Half of the small bowel appeared dilated and ischemic, the other half empty and placed in an unusual location: we found a mesenteric breach where the transverse colon and the empty part of the small bowel appeared strangulated. The small bowel was freed from the breach, untwisted and replaced on the right side of the abdominal cavity. The vitality of the whole bowel was tested and positively confirmed. Due to the emergency setting and the severe dilation of the cecum, the malrotation was left uncorrected and the cecum wasn't fixed to the abdominal wall. The post-operative recovery was uneventful and the patient was discharged on 4th POD.

The follow-up at one, three and five months was characterized by isolated episodes of subocclusion with no need for hospitalization. During the follow-up period we performed an upper gastrointestinal contrast study and a CT-scan with water-soluble contrast that confirmed the reverse midgut rotation.

The patient at the time refuses the definitive correction of his condition.

Discussion

Beckwith-Wiedemann syndrome is a rare genetic disorder caused by the alteration of growth-regulating genes of the chromosome region 11p 15.5. The incidence of this condition is approximately 1:10.000 live births [6,7].

Its manifestations are variable, including postnatal overgrowth, hypotonia, nevus flammeus of the forehead, macroglossia, prognathism, pinna abnormalities, heart anomalies, supernumerary nipples, abdominal wall defects (omphalocele, umbilical hernia, diastasis recti), visceromegaly (liver, pancreas, spleen or kidneys), malformations of the kidney and ureter, whole-body hemihypertrophy, hyperinsulinemic hypoglycemia and increased risk of embriogenic tumors (hepatoblastoma, Wilms' tumor) [8,5,6].

Our patient was born with the typical presentation of the BWS with: macroglossia, umbilical hernia, and hypoglycemia. The patient didn't report any abdominal symptoms since three years before the event.

Intestinal malrotation is a rare disorder, more commonly diagnosed during infancy and childhood, even if it may lead to disastrous ischemic consequences at all ages. The incidence of malrotation is 1:6000 live births according to post-mortem studies [9].

The embriological midgut is composed of two portions that have different functions and developement during embriological life. The cephalad pre-arterial portion gives origin to the distal duodenum, jejunum and proximal ileum while the caudad one develops into distal ileum, cecum, appendix and colon [12].

A complete development is obtained with a 270° counterclockwise rotation around the axis of SMA[9] and occurs in different stages. The first includes the herniation of the midgut outside of the abdominal wall and its 90° counterclockwise rotation. The cephalad midgut goes downward, right to SMA, while the caudad one locates upward on the left of SMA. The duodenojejunal loop undergoes another 90° counterclockwise rotation, and as the abdominal cavity develops the physiological umbilical hernia regresses. After 10 weeks, the duodenojejunal loop complete a final 90° counterclockwise rotation and the third duodenal segment places inferiorly and posteriorly to the SMA and then upward to the left of the artery creating the Treitz ligament. The caudad portion undergoes an additional 180° counterclockwise rotation, finally locating anteriorly to the SMA with the cecum placing in the right iliac fossa. The last stage is the fixation of the mesentery to the posterior abdominal wall [14].

Different types of intestinal malrotation are currently known, depending on the involved stage of midgut developement: non-rotation, incomplete rotation, mixed rotation and reverse rotation [12,13].

The reverse rotation is the most rare malrotation, accounting for 2-4% of all cases and it is more common in women [15]. It origins from a premature return of the caudad midgut into the abdominal cavity while the duodenum loop rotates clockwise. The cecum begins its migration and shifts to the right behind the SMA. As a result, the transverse colon lies behind the duodenum and is separated from it by the SMA. This prevents the formation of Treitz ligament [14].

Due to the rarity of this condition, little is known about the real indicence of complications. The impaired fixation and subsequent excessive mobility of the ascending colon and cecum imply an increased risk of developing an ileo-cecal volvulus. Other complications observed in reverse midgut rotation are obstruction of the transverse colon by the SMA, and duodenenojejunal obstruction due to paraduodenal herniation or bands. These situations require rapid diagnosis and surgical management. When intestinal malrotation is suspected, the most reliable radiologic sign is the intraperitoneal location of the third duodenal segment, just as inverted mesenteric vessels [13].

In literature few studies describe an association between midgut volvulus and abdominal wall defects as the omphalocele, but no one describes the association between BWS and intestinal malrotation [10]. There is only one study that report the strong relation between syndromic conditions and genetic disorders, however BWS isn't mentioned [11].

Surgery is the gold standard in the management of acute complications resulting from intestinal malrotations. The standard of care consists in Ladd's procedure: surgical division of bands and of possible adhesions near the SMA, counterclockwise rotation of the bowel, in order to relieve the obstruction and widen the base of the

mesentery to prevent future volvulus, associated to a prophylactic appendectomy. There are other surgical procedures described to correct the rotation, like right hemicolectomy or resection and displacement of transverse colon anteriorly to the mesenteric root [13].
The adequate treatment of asymptomatic patients with incidental diagnosis of malrotation instead is still matter of debate. The majority of pediatric surgeons recommend a surgical correction in any given case, but there is still no evidence for or against surgery versus a watchful waiting approach [13].

Conclusion

Intestinal malrotation is an uncommon disease that may interest also adult population, often presenting with acute symptoms that require immediate recognition and surgical approach. Our case described a syndromic adult patient with small and large bowel obstruction due to jejunal transmesenteric internal hernia in reverse midgut rotation. Emergency surgery is the standard of care for acute complications of this rare condition, so every General Surgeon should be prepared to recognize this uncommon complication during the first hours of presentation.

Riassunto

La malrotazione intestinale rappresenta una rara condizione congenita con un'incidenza nel paziente adulto che a seconda delle casistiche varia circa tra lo 0.0001% e lo 0.19%, colpendo 1:500 neonati. Questa rara condizione è causata da un'anomala rotazione dell'intestino all'interno della cavità peritoneale durante l'embriogenesi. Nella maggior parte dei casi coinvolge sia l'intestino tenue che il colon, causando un elevato rischio nel corso della vita dell'individuo affetto di andare incontro ad episodi di occlusione intestinale.
Esistono un'ampia varietà di anomalie anatomiche causate dal blocco della rotazione della porzione media dell'intestino ed esse dipendono dalla fase di sviluppo embriologico in cui tale blocco si presenta. La rotazione inversa è la forma più rara di malrotazione intestinale (2-4% del totale) ed è più comune in individui di sesso femminile. Tale anomalia origina da un ritorno prematuro della parte caudale della porzione media dell'intestino nella cavità addominale, nel momento della vita fetale in cui il duodeno ruota in senso orario, ovvero fra la quarta e la dodicesima settimana gestazionale. Il cieco in quel momento inizia la sua migrazione verso la fossa iliaca destra passando quindi posteriormente all'arteria mesenterica superiore anziché anteriormente. Il risultato è quindi che il colon si posiziona posteriormente all'arteria mesenterica superiore e anche al duodeno.
Le più comuni manifestazioni cliniche della malrotazione intestinale nel neonato sono l'occlusione intestinale alta a livello del duodeno e il volvolo della porzione media dell'intestino, condizioni che possono mettere a rischio la vita del neonato progredendo verso l'ischemia intestinale acuta. Nell'individuo adulto il rischio di volvolo diminuisce e la presentazione clinica risulta più aspecifica, conducendo a difficoltà e conseguenti ritardi diagnostici che possono avere conseguenze disastrose per il paziente.
Riportiamo un raro caso di maschio adulto presentatosi per la prima volta alla nostra attenzione con sintomatologia acuta di occlusione intestinale causata da una malrotazione intestinale, nello specifico da rotazione inversa, nel contesto di una sindrome di Beckwitt-Weideman, un raro disordine genetico caratterizzato dalla compresenza di citomegalia surrenalica, emipertrofia, macroglossia, onfalocele e iperplasia delle isole pancreatiche.

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