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Case Report

ISOLATED ANORECTAL MALFORMATION IN TWO SIBLINGS AS CAUSE OF CHRONIC CONSTIPATION. CASE REPORT

MALFORMACIÓN ANORRECTAL AISLADA EN DOS HERMANOS COMO CAUSA DE CONSTIPACIÓN CRÓNICA. REPORTE DE UN CASO

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Abstract

Anorrectal malformations (ARM) are a broad spectrum of congenital malformations and a challenge for surgeons. These can be part of a syndromic presentation or sporadic / familial, but there are few reports of ARM in relatives of first degree of consanguinity. Chronic constipation not responsive to laxatives has been described as one of the most frequent symptoms leading to the diagnosis of isolated and inadvertent ARM in pediatrics, which increases morbidity and mortality.

Two siblings, male and female, with different forms of presentation and late diagnosis of isolated ARM with rectoperineal fistula that required posterior sagittal anorectoplasty (PSARP). Both patients had satisfactory recuperation and good fecal continence.

Having isolated ARM with rectoperineal fistula in two siblings is a rare form of presentation. The rate of ARM recurrence among siblings and relatives remains unknown given the limited literature reported. The late diagnosis of ARM implies high mortality and morbidity in the short and long term for fecal continence.

In pediatric patients with severe chronic constipation not responsive to laxative, it is mandatory to rule out ARM. The diagnosis could be a challenge in cases of isolated non-syndromic ARM and eventually delay the surgical management, thus increasing the morbidity and mortality of these patients. In patients with ARM with perineal or vestibular fistula, multidisciplinary and genetics consultation should be carried out to rule out recurrence between siblings.

Resumen

Las malformaciones anorrectales (MAR) son un amplio espectro de malformaciones congénitas y un reto para los cirujanos. Estas pueden ser parte de una presentación sindrómica o esporádica/ familiar, pero existen pocos reportes de MAR en familiares de primer grado de consanguinidad. El estreñimiento crónico que no responde a los laxantes se ha descrito como uno de los síntomas más frecuentes que conducen al diagnóstico de MAR aislada e inadvertida en pediatría, lo que aumenta la morbimortalidad.

Dos hermanos, masculino y femenino, con diferentes formas de presentación y diagnóstico tardío de MAR aislada con fístula recto perineal que requirió Anorrectoplastia sagital posterior (PSARP). Ambos pacientes tuvieron una recuperación satisfactoria y buena continencia fecal.

Tener una MAR aislada con fístula recto perineal en dos hermanos es una forma de presentación rara de esta anomalía. La tasa de recurrencia de MAR entre hermanos y familiares sigue siendo desconocida debido a la literatura limitada reportada. El diagnóstico tardío de las MAR implica una alta mortalidad y morbilidad a corto y largo plazo por la continencia fecal.

En pacientes pediátricos con estreñimiento crónico severo que no responden a laxantes, es obligatorio descartar MAR. El diagnóstico puede ser un desafío en casos de MAR no sindrómica aislada y eventualmente retrasar el manejo quirúrgico, aumentando así la morbimortalidad de estos pacientes. En pacientes con MAR con fístula perineal o vestibular se debe realizar consulta multidisciplinar y de genética para descartar recurrencia entre hermanos.

Keywords: anorectal malformation, constipation, colorectal surgery, congenital malformations, inheritance, ARM.

Introducción

Anorectal malformations (ARM) are a broad spectrum of congenital malformations that affects colorectal health in pediatric population. The incidence rate reported is 1:1,500 to 1:5,000 live births (1–4). Despite of been widely studied, this group of anomalies continues to be challenging for surgeons because of its clinical presentation (2,5) Although the cause of ARM is unknown, defects in determinants during blastogenesis have been implicated (6).

ARM can be classified according to their clinical management, postoperative outcome or location. From the genetic spectrum, they can be divided into non-syndromic (ARM as the only pathological finding) with different inheritance patterns, and syndromic ARM, reported in 60% of cases (7–9), associated with a complex phenotype (Townes-Brocks syndrome, Currarino syndrome, FG syndrome, VACTERL, among others) (2–5). However, some authors report a low rate of association in families, unlike others who report an autosomal dominant pattern of inheritance (2).

ARM occur because of abnormalities in the development of the hindgut, which is the embryological structure that forms the descending colon, rectum and the upper part of the anal canal, the lining of bladder, and the urethra. The hindgut enters the posterior region of fetal cloaca (cavity of endoderm and ectoderm), in which the hindgut and allantois converge. During the 7th week of development, the cloacal membrane separates, creating the ventral opening for urogenital sinus (urinary tract) and the anal opening for the hindgut, with the perineal body forming between the two, explaining the variety of anorectal defects. (4–6).

Knowing the embryological development, its mandatory that all newborns receive a general examination, including the perineum, evaluating how many orifices are in the perineum, the absence or characteristics of the anus, the anal fossa, the genitalia, the urethra, the presence of meconium and the permeability of the anus with a catheter (2,3,10). According to findings in physical exam, pediatricians, neonatologists, and pediatric surgeons decide to take imaging studies to define treatment and surgical management of patients with ARM and define fecal continence prognosis, all within the first 72 hours of newborns.(1)

Severe chronic constipation not responsive to laxatives and diet has been one of the most frequent symptoms leading to the diagnosis of isolated and incidental ARM in pediatric patients, which increases morbidity and mortality (8,11–13). Patients with ARM do not have a normal anal canal, have variable sphincter deficiencies, bowel motility disorders, which predisposes them to high morbidity despite having ARM with a good continence prognosis (14).

Cases presentation

We present a case report of two siblings, male and female, with late diagnosis of isolated ARM with rectoperineal fistula. We made the diagnosis of one of them incidentally. Both siblings needed surgical management with posterior sagittal anorectoplasty (PSARP).

Case 1

A 2-year-old female patient without history of any medical problem was admitted to the emergency department referred by pediatric gastroenterology due to abdominal pain, vomiting and no stools for 8 days. Additionally, with a history of chronic constipation and multiple hospital admissions because of fecal impaction, apparently due to Hirschsprung disease. Also, the patient had a report of an extra institutional barium enema with an apparent transition zone in the descending colon, which made pediatricians considered Hirschsprung disease as diagnosis and started rectal irrigations. After this, the patient was evaluated by Pediatric Surgery department because of persistent abdominal pain, distension and palpation of fecalomas, with evidence of an anterior anus. Report of abdominal X-ray with dilatation of small bowel loops and colon. However, given poor clinical evolution and physical exam, we decided to do an anal examination with electrocutaneus stimulation of the anus and perineum under general anesthesia, showing an anterior anus, differences in color of skin of the perineum and anus; also, with electrostimulation we observed posterior contraction of the annal sphincter but with the anal opening located outside of the center of the sphincter. We diagnosed anorectal malformation with rectoperineal fistula (Figure 1). Because of these findings and fecal impactation we performed a Hartmann-type colostomy and later a PSARP.

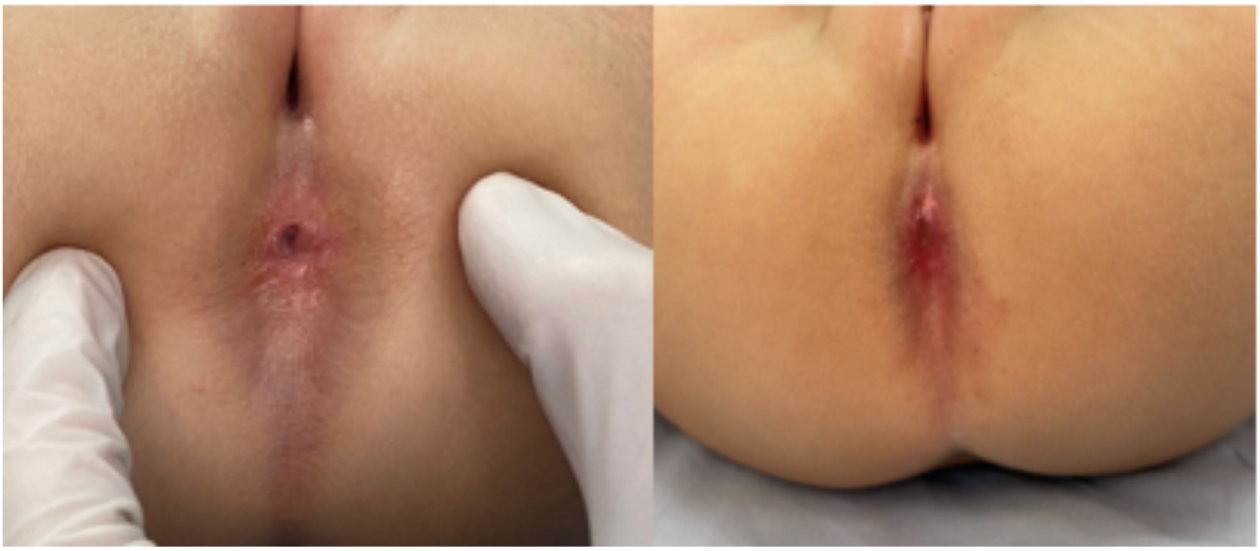

Figure 1. Findings in anal examination with electrostimulation under general anesthesia: Anorectal malformation with recto-perineal fistula.

Surgical technique: PSARP

Verification of informed consents, surgical safety checklist, under general anesthesia. Placement of Foley catheter (Num. 8). In prone slightly jack-knifed position (with elevation of the hip), asepsis and antisepsis of the surgical field.

First, we identified the position of the muscular complex with an anal and perineum electro stimulator, then we placed circumferential traction sutures with 3-0 silk suture. We made the incision in the midline towards the coccyx and made the dissection of the fistula and rectum with monopolar energy until identification of elevator ani muscle, always observing the white fascia of the rectum to avoid opening of the posterior aspect of the mucosa. Later, we identified parasagittal fibers with electrostimulator, showing the anterior and posterior limits of the muscle complex and marked the limits with 3-0 silk sutures. We continued the dissection of the rectum until reaching adequate length of the rectum for mobilization, then we calibrated the position of the rectum in the center of the muscular complex. Closure of perineal body with multiple layers of separate absorbable sutures and closed the posterior aspect of the muscle complex. We sectioned opened the rectum and the rectal fistulous tract until seeing an adequate caliber of the neoanus. Then we closed the anterior aspect of the muscle complex with 3-0 absorbable sutures and seccioned the colon in half and sequentially anastomosed it to the skin with sixteen absorbable sutures placed through the bowel and skin and completed the anoplasty making the skin closure with absorbable suture. Finally, we passed hegar dilator into the neoanus until reaching number 12 without resistance and checked adequate position of the neoanus and muscle complex with electrostimulator. (Figure 2)

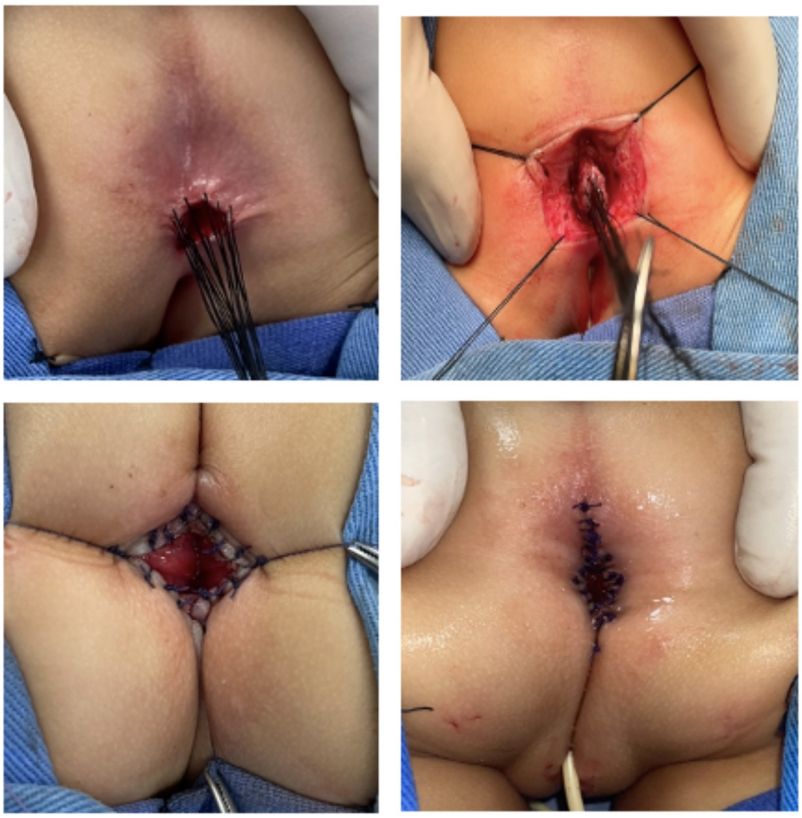

Figure 2. Intraoperative images. PSARP

Case 2

A 5-year-old male patient admitted to pediatric surgery consult, with a history of a sister with ARM with perineal fistula and chronic constipation managed with diet and polyethylene glycol (PEG) with poor improvement, semi-hard stools every 3-4 days. Physical exam with mild abdominal distension and perineum with an ARM with perineal fistula and bucket handle sign (Figure 3.). We requested a barium enema with finding of dolichosigmoid. Because of this, we performed a PSARP without diverting colostomy. (Figure 4.)

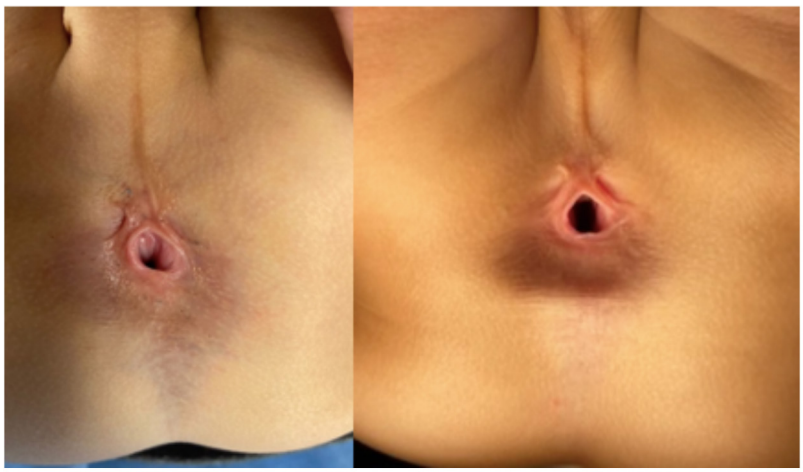

Figure 3. Perineum. ARM with perineal fistula and bucket handle sing, slightly flattening of the inter- gluteal cleft, anterior anus.

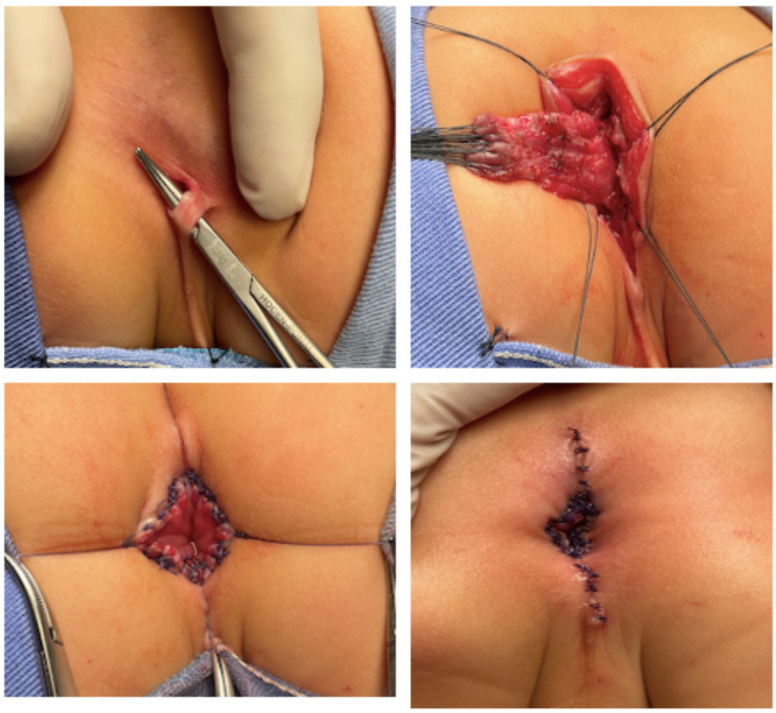

Figure 4. Intraoperative images of PSARP

Results

Both patients had satisfactory recuperation, stooling every day and fecal continence.

Case 1: The patient followed postoperative dilations and reached the indicated dilator according to her age (Hegar 15 to 16). Six months after the dilations, we performed the colostomy closure with excellent postoperative results, stooling, and fecal continence.

Case 2: Satisfactory postoperative results, with positive bowel habits with soft stools 1-2 times a day, with flatus and fecal continence, without accidents or fecal soiling.

Discussion

ARM present as a spectrum of variable defects affecting urogenital and gastrointestinal tract. Some of them are malformations that with a successful surgical reconstruction have excellent results for bowel, urinary and sexual function; but, some of them are complex and devastating malformations with anatomic deficiencies that repercuss and affect patients life in short and long term (4,5).

Traditionally, ARM were classified into three different categories (“high”, “intermediate” and “low), until 1980 when Alberto Peña created a new classification based on therapeutic and prognostic factors leading surgeons to know the specific treatment and functional prognosis of each malformation (4,15). Also, in 1982, Peña described the PSARP as the best surgical approach to observe the intrinsic anatomy of the pelvis and manage the anomalies associated with ARM, with a strong impact in the quality-of-life of patients and their aftercare (16,17), and with more than 3,000 patients follow up and results, Peña´s PSARP became the best surgical approach for the management of ARM regardless of the fistula location or complexity of the malformation. (3,4,16,18,19)

As colorectal pediatric surgeons reported, ARM with rectoperineal fistula and ARM without fistula (in male or female) are the malformations with best prognosis for fecal continence (4,5,20), like our case, in which both patients presented ARM with rectoperineal fistula and after surgery they have good fecal continence, adequate stooling and bowel motility.

On the other hand, having isolated ARM (without other pathological finding) with rectoperineal fistula in two siblings is a rare form of presentation. There are few reports in literature that describe the existence

of ARM in relatives of first degree of consanguinity. Anderson reported a 1% risk of recurrence, concluding that isolated ARMs are a rare entity (7). In 2005, Kubiak published a review of the literature on cases of families with two or more siblings with isolated ARM, showing that there were only 19 families with a diagnosis of ARM, but with different location, phenotype, or little information on the characteristics of the malformation (21). Also, they reported a case of monozygotic twins with ARM without fistula. (22)

Furthermore, Scharamm et al. reported a case of a mother and her two children with ARM in whom a genetic study was performed without finding the causal genetic alteration. Also, they reported a review that showed 59 families with at least two members affected with ARM. (13)

Dworschak et al. published a cohort of 619 patients with ARM where they found that the risk of recurrence for the offspring of a patient with ARM was increased 32 times if one sibling was affected (7). In the same way, Falcone et al., reported finding that patients diagnosed with ARM with perineal or vestibular fistula have a higher risk of having a relative with ARM (23). This finding, in addition to our case report, helps pediatric surgeons suspect and associate the risk of recurrence in patients with ARM and perineal fistula among siblings and evaluate the risks in their offspring.

On the other hand, in patients with chronic constipation it is always necessary to evaluate the presence of ARM as it can go unnoticed in neonatal period and could be associated with complications that increase morbidity and mortality in patients with ARM (24). Fecal impaction, megacolon, pseudo incontinence, colostomy requirement, among others, are part of the complications associated with patients with ARM, causing a delay in performing the definitive surgical procedure (24–26).

There aren't case reports in literature of two siblings with incidental finding of ARM with perineal fistula as cause of chronic constipation, as was shown in this case report. Some authors have reported specific chromosome and gene mutations associated with ARM. But, given of our healthcare system limitations, it was not possible to stablish a genetic cause or gene mutation to explain the presentation of this malformation in two siblings.

In summary, the rate of ARM recurrence among siblings and relatives remains unknown given the limited literature reported. The late diagnosis of ARM implies high mortality and morbidity in short and long term for patients (4,5,25,26). For this reason, a case of two siblings, a boy, and a girl, with late diagnosis (at 2 and 5 years of age, respectively) of ARM with isolated perineal fistula is presented. With this case, pediatricians, neonatologist and healthcare workers can be aware of the emotional, economic and surgical implications and impact that arise from delayed diagnosis and late consultation to pediatric surgery in pediatric patients with chronic constipation. Also, the repercussions of this complex malformations in adult and elderly life without early diagnosis and management.

Conclusions

ARM continue to be a challenge for surgeons. In pediatric patients with severe chronic constipation not responsive to laxative, it is mandatory to rule out ARM. The diagnosis could be a challenge in cases of isolated non-syndromic ARM and eventually delay the surgical management, thus increasing the morbidity and mortality of these patients. In patients with ARM with perineal or vestibular fistula, multidisciplinary and family consultation should be carried out to rule out recurrence between siblings.

Competing interests:

The authors declare no conflicts of interest and report that there are no competing interests either. Furthermore, the authors followed institutional and ethical protocols and did not obtain funding for this work.

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