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RESEARCH

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Overlooked anterior anal misplacement: a 'forme fruste' anomaly and potentially correctable cause of constipation

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Abstract

Purpose To share our experience in the management of a specific type of mild anorectal anomaly presenting with severe constipation.

Methods Data of patients with idiopathic constipation who were referred to our pediatric surgical facility during the period from 2013 to 2023 were retrospectively analyzed. The study included patients with significant symptoms of constipation associated with a minor degree of unrecognized anterior anal misplacement. MRI was ordered in most cases to confirm the presence of deeper structural abnormalities.

Results Data from 16 cases were available for retrospective analysis. All included cases underwent posterior anoplasty with posterior sphincterotomy. Postoperative recovery was uneventful in all cases with no wound complications. Almost all cases reported obvious improvement of symptoms regarding frequency of defecation and relief of straining which was the most distressing preoperative complaint. After 1 month, 12 cases were off laxatives with regular defecation; 3 cases still required lower doses of laxatives. Only one case reported unsatisfactory response up to 3 months postoperatively, and unfortunately was lost to follow-up. Five cases responded to messages sent to them through registered phone numbers and were available for longer-term follow-up.

Conclusion Mild unrecognized forms of anorectal anomalies may present with idiopathic constipation. Severe straining at defecation is an important clue. The subtle abnormality on perineal examination can be supported by pelvic MRI unveiling more structural abnormalities at a deeper level. Posterior anoplasty offered relief of symptoms in most cases.

Keywords Ectopic anus, Perineal fistula, MRI, Anal canal, Defecation

Background

There are two main surgical causes for constipation in children that are essentially managed by pediatric surgeons: Hirschsprung disease and anorectal anomalies. Habitual or idiopathic constipation is a broad term used when no apparent cause for constipation is found [1, 2]. It is not uncommon for a pediatric surgeon to be involved in managing cases of habitual constipation as well. One important task is to search for unrecognized surgical causes among these cases when they can benefit from a surgical solution for their chronic problem [1, 2].

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There are few reports in the literature that referred to the presence of slight unrecognized anterior anal misplacement among children (mostly girls) suffering from chronic constipation with severe straining at defecation. In the same year 1978, and in two separate reports, Hendren [3] and Leape and Ramenofesky [4] described children presenting with constipation associated with slight anterior anal displacement who were successfully treated by posterior anoplasty. The diagnosis can be initially missed due to minimal displacement of the anus when it may be considered a normal individual variation in the topography of the perineum [3]. Often, these cases were referred with a diagnosis of suspected Hirschsprung disease [3]. Later in 1984, Upadhyaya reported a similar observation in a smaller case series, where he tried to provide explanation for constipation and severe straining in this group of children with mild anterior displacement of the anus based on his clinical and operative findings [5].

In our practice, we have encountered similar cases with the typical presentation of severe straining at defecation associated with subtle deviation of the external anal position that was successfully managed by posterior anoplasty. Here, we would like to add this new case series supported by pelvic MRI studies demonstrating deeper anatomical aberrations in this mild frequently unrecognized form of the anomaly.

Methods

Data retained by the author for patients with idiopathic constipation who were referred to our pediatric surgical facility during the period from 2013 to 2023 were retrospectively analyzed. The study included patients with significant symptoms of constipation and in whom we suspected a minor degree of unrecognized anterior anal misplacement. The criteria (inclusion and exclusion criteria) to select such cases with debatable diagnoses can be summarized in the following points:

First, the history of constipation in these children was usually dated since early infancy being more pronounced after the start of weaning. Remarkably, almost all cases passed stools with severe straining whatever the frequency of defecation (sometimes up to every 5–10 days); otherwise, they were healthy children. Some parents reported that they used to press on the perineum of their children behind or at both sides of the anus to facilitate the extrusion of feces through the anus (Fig. 1).

Second, on examination, the anus looked 'more or less' normal with no evidence of stenosis. However, in females (majority of cases), the perineum looked shorter than usual (Fig. 2). With more careful examination, one can notice the peri-anal pigmented skin is not evenly distributed around the anus. In other words, the anus was not perfectly in the middle but slightly anteriorly displaced (eccentric), which may still be considered normal by many of the attending pediatric surgical colleagues due to the absence of stenosis. Occasionally, an anterior anal fissure was noticed at the 12 o'clock position (Fig. 2).

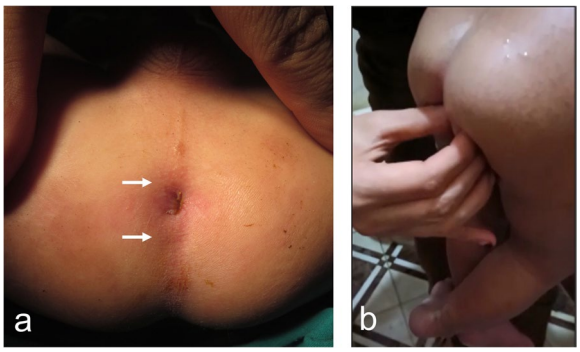

Fig. 1 Fifteen-month-old boy presenting with constipation characterized by severe straining at defecation not responding to medical treatment. **a** Photograph of the perineum in the supine lithotomy position: the anus was not perfectly in the middle, but slightly anteriorly displaced with no evidence of stenosis. White arrows mark the anterior and posterior boundaries of the perianal pigmented skin. **b** Photograph sent by the mother to demonstrate how she facilitates the extrusion of stools by squeezing on the perineum at both sides of the anus

Third, it was important to exclude the possibility of Hirschsprung disease in these patients. The typical history of Hirschsprung disease includes delayed passage of meconium in addition to significant abdominal distension which is considered the red flag for diagnosing Hirschsprung disease. Failure to thrive and enterocolitis are another two important associations with Hirschsprung disease. Cases included in the study lacked this typical history and associations. In addition, their contrast enema demonstrated a normal/dilated rectum down to the anal canal usually with prominent posterior shelving (Fig. 3). In doubtful situations, a rectal biopsy was performed that confirmed the presence of ganglion cells. In some cases, rectal biopsies were already taken before referral, which excluded Hirschsprung disease.

Fourth, to support the subtle external deviations, pelvic MRI studies were ordered in most cases searching for possible deviations from the norm at a deeper level. Findings were related to our previous experience comparing pelvic MRI anatomy in cases of anorectal anomalies and controls [6, 7]. Interestingly, we could detect deviations similar to those found in mild forms of anorectal anomalies (rectoperineal fistula) [8]. These mild deviations from the norm were more pronounced at the level of the mid-anal canal rather than distally. The mid-anal canal appeared elongated (Fig. 4), hanging forwards, and anteriorly displaced in relation to bony and soft tissue

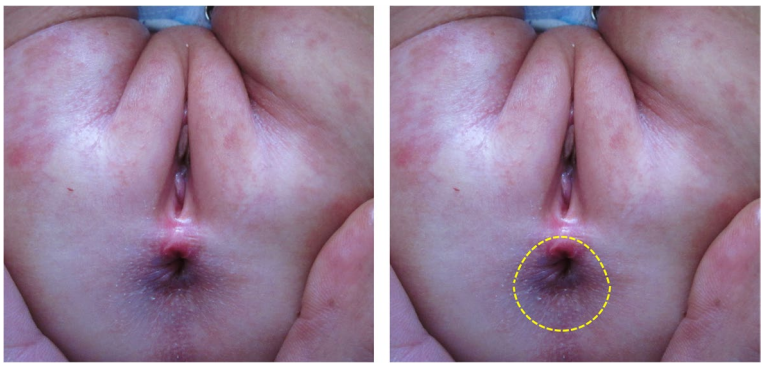

Fig. 2 Eight-month-old girl presenting with constipation characterized by severe straining at defecation. Photograph of the perineum in the supine lithotomy position: notice the peri-anal pigmented skin is not evenly distributed around the anus (dotted yellow circle); the anus is not perfectly in the middle but slightly anteriorly displaced with no evidence of stenosis. Note the presence of an anal fissure at the 12 o'clock position

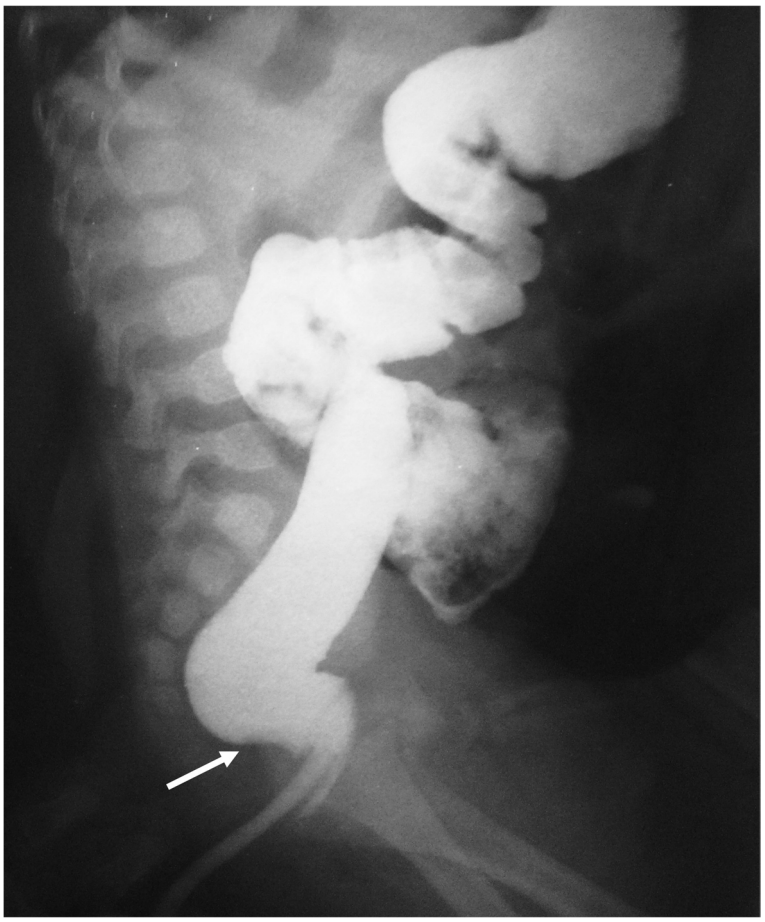

Fig. 3 Contrast enema demonstrating normal rectum down to the anal canal with prominent posterior shelving (arrow)

landmarks: ischial tuberosities (Fig. 5), and transverse perineal muscles (Fig. 6).

By collecting the above pieces of evidence, and most importantly, in the presence of the characteristic complaint of severe straining during defecation (indicating some sort of mechanical obstruction), we offered these cases surgery in the form of posterior anoplasty [3].

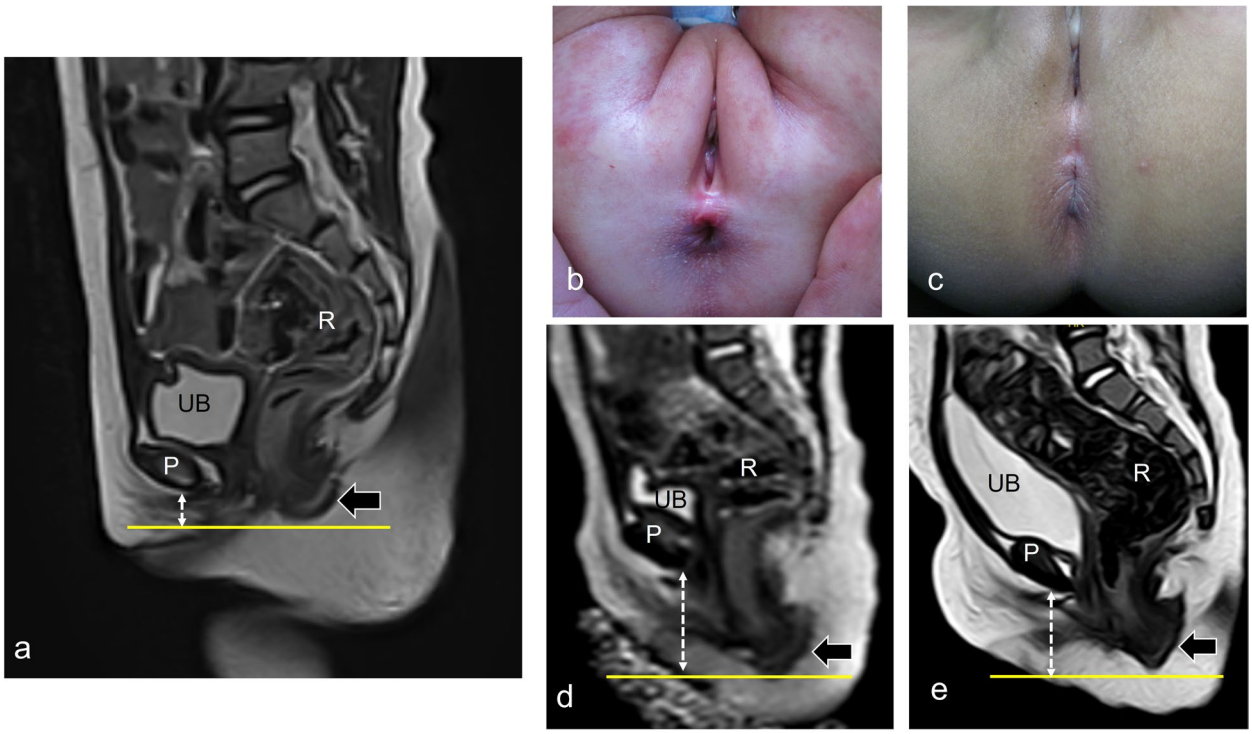

Fig. 4 Deep pelvic anatomical deviations in cases with mild anterior anal misplacement as demonstrated in their pelvic MRI studies compared to normal control (**a**). **a** Mid sagittal pelvic MRI (T2WI) in 5-year-old girl (normal control). **b**, **c** Eight-month-old and 2-year-old girls with mild anterior anal misplacement presenting with severe constipation. **d**, **e** Mid sagittal pelvic MRI (T2WI) of the above two cases (**b** and **c**, respectively). P: pubic symphysis; UB: urinary bladder; R: rectum; thick black arrow points to the site of the anus. Note the elongated anal canal in **d** and **e** compared to control (**a**); a double arrowhead dotted line is used to mark the distance from a fixed bony landmark, the pubic symphysis (P)

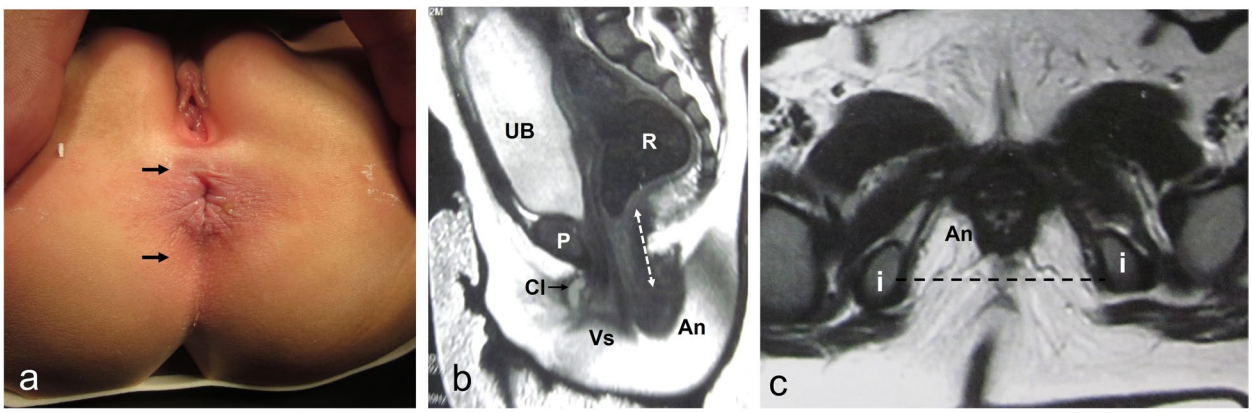

Fig. 5 One-year-old girl with mild anterior anal misplacement presenting with severe constipation. **a** Photograph of the perineum in the supine lithotomy position: the anus was not perfectly in the middle, but slightly anteriorly displaced with no evidence of stenosis. Arrows are marking for the anterior and posterior boundaries of the perianal pigmented skin. **b**, **c** Pelvic MRI study mid-sagittal, and axial (at the level of ischial tuberosities), respectively, demonstrating deep pelvic anatomical deviations. P: pubic symphysis; UB: urinary bladder; R: rectum; An: anus; Vs: vestibule; Cl: clitoris; i: ischial tuberosity. Note the distended rectum (R) and elongated mid portion of the anal canal in **b**. Also, note the anterior displacement of the anal canal (An) in **c**, which should be normally located at the level of an imaginary line joining both ischial tuberosities (i)

Surgical technique (posterior anoplasty)

The patient is placed in the supine lithotomy position (Fig. 7a). A posterior circum-anal incision is made from the 3 o'clock to the 9 o'clock position. The incision is deepened by dissection on the posterior and lateral aspects of the anal canal; this step is facilitated

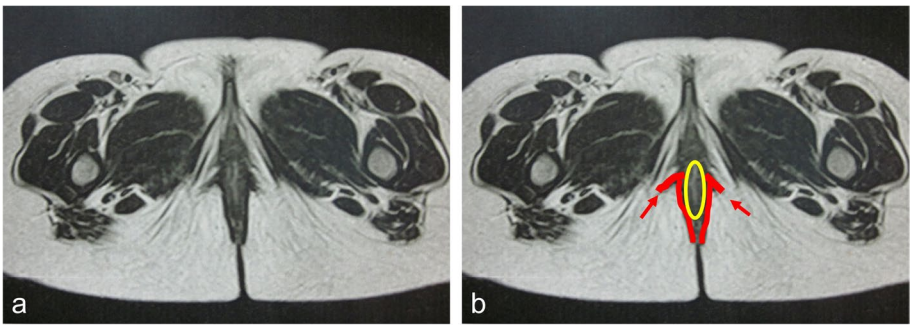

Fig. 6 Seven-month-old girl with mild anterior anal misplacement. **a** Axial pelvic MRI (T2WI) at the level of the distal anal canal with color-overlay in **b**. Note the anal canal (yellow circle) is displaced anteriorly separating the transverse perineal muscles (red arrows) on both sides

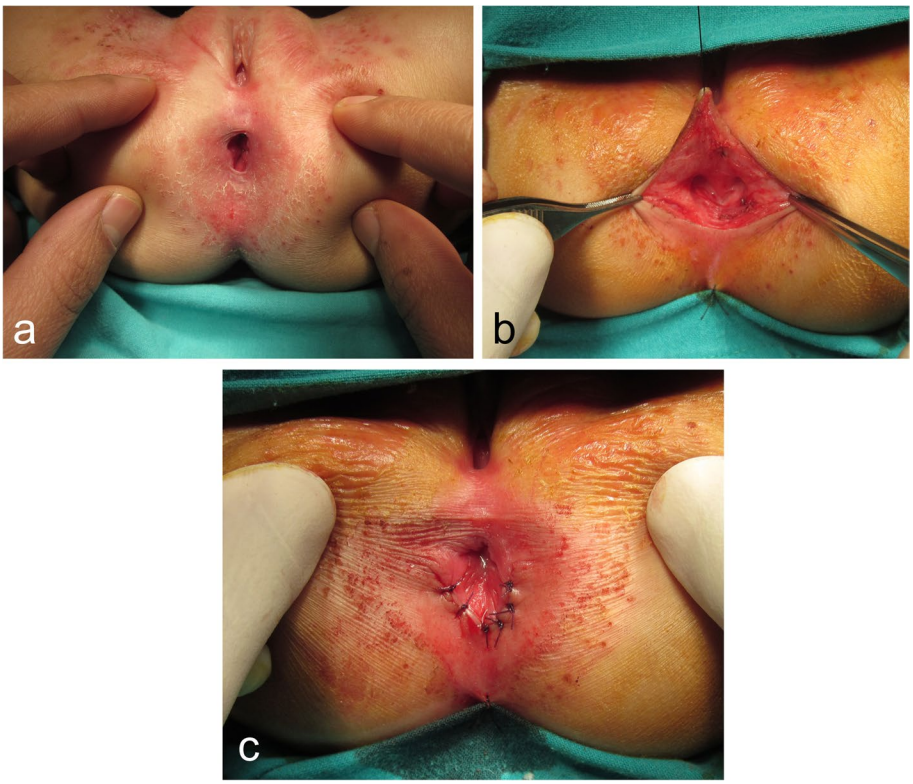

Fig. 7 Surgical technique of the posterior anoplasty. **a** The patient is placed in the supine lithotomy position. **b** A posterior circum-anal incision is made from the 3 o'clock to the 9 o'clock position. **c** After performing the posterior myotomy, the skin incision is then closed

by outward retraction of the anus (Fig. 7b). The posterior traversing subcutaneous sphincteric muscle fibers are identified deep in the wound, which are incised in the midline (posterior myotomy). The skin incision is then closed (Fig. 7c). More recently, we modified the procedure to be performed in a simpler yet similar way (Fig. 8). A vertical midline "cutback" incision (about 1 cm) is made in the posterior wall of the anal canal and nearby skin (at the 6 o'clock 'position) (Fig. 8a). The posterior extension of this vertical 'cutback' incision is limited by the boundaries of the modified (pigmented) perianal skin. Deep in the wound, the posterior traversing muscle fibers are identified and incised in the midline (posterior myotomy) (Fig. 8e). The vertical mucocutaneous incision is then closed in a horizontal fashion. Puckering of the skin/mucous membrane at the edges of the wound may need to be trimmed (Fig. 8).

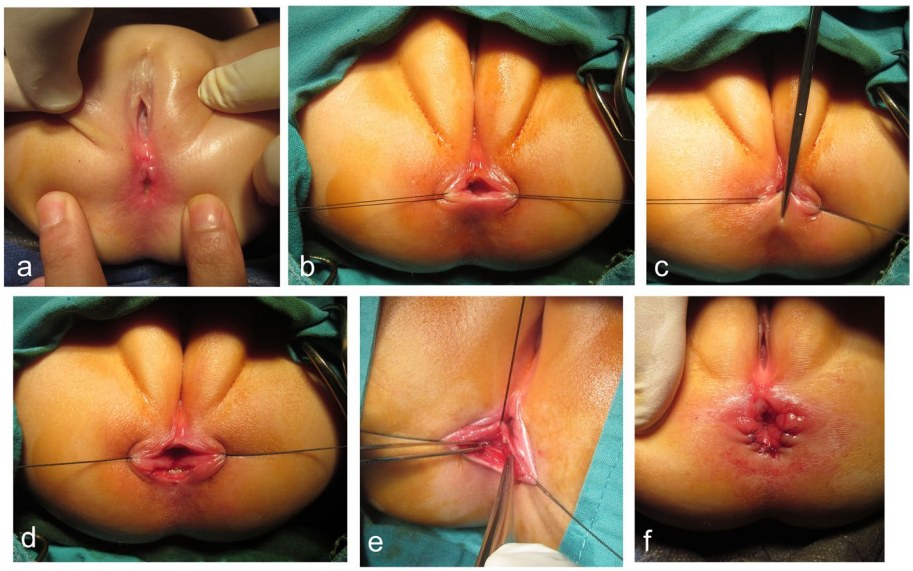

Fig. 8 Surgical technique of the cutback anoplasty. **a** The patient is placed in the supine lithotomy position. **b** Traction sutures at the 5 and 7 o'clock positions. **c**, and **d** A vertical midline "cutback" incision (about 1 cm) is made in the posterior wall of the anal canal and nearby skin (at the 6 o'clock 'position). **e** Deep in the wound, the posterior traversing muscle fibers are identified and incised in the midline (posterior myotomy). **f** The vertical mucocutaneous incision is then closed in a horizontal fashion

Postoperative care

Enteral feeding is resumed on the same day upon full recovery, and the patient is discharged home the next day on analgesics. No need for prophylactic antibiotics apart from a single dose given at induction of anesthesia. Laxatives are usually prescribed on the third postoperative day and continued for 2–3 weeks as needed.

Follow-up

The first follow-up visit was scheduled 7 days postoperatively to inspect for possible wound complications. As the incision was not involving the whole anal circumference, postoperative anal dilatation was not required. The second visit was 4 weeks later to ask about symptoms of constipation (straining), frequency of defecation, and whether there was a need to continue on laxatives. Messages were sent to parents of operated cases through their registered phone numbers to ask about longer-term follow-up.

Results

Data from 16 cases that fulfilled the above-mentioned inclusion criteria were available for retrospective analysis. Their age upon referral ranged between 7 and 60 months (mean = 23, median = 16). Most of the cases were girls (14 cases, 87.5%). Anterior anal fissure (12 o'clock position) was observed in three girls. Preoperative MRI studies were performed in 13 cases (81%), which confirmed the presence of the above-mentioned deviations from the norm but with variable degrees (Figs. 4, 5, and 6). Before referral, all cases received medical treatment for constipation (laxatives). In most cases, laxatives were ineffective in relieving symptoms especially those related to severe straining at defecation. In only one case, the parents reported a satisfactory response to laxatives; however, symptoms recurred each time they stopped the medical treatment.

All included cases underwent posterior/cutback anoplasty with posterior sphincterotomy as described in the methods. Postoperative recovery was uneventful in all cases with no wound complications. Within 2–4 weeks postoperatively, almost all cases reported obvious improvement of symptoms regarding frequency of defecation and relief of straining which was the most distressing preoperative complaint. After 1 month, 12 cases were off laxatives with regular defecation (at least 4 times/week); 3 cases still required lower doses of laxatives. Only one case reported unsatisfactory response up to 3 months postoperatively and unfortunately was lost to follow-up. Five cases responded to messages sent to them through registered phone numbers and were available for longer-term follow-up. The follow-up period ranged between 2 and 7 years (mean = 4 years $\pm$ 2, median = 3 years). When asked about the effect of the operation, parents showed great appreciation reporting relief of straining/suffering compared to the preoperative condition; no one complained of postoperative faecal incontinence. Parents of three cases reported that their children were

cured (no laxatives) and were doing well with no need to seek further medical advice. The other two cases reported occasional constipation that required returning to laxatives for short periods; however, parents were generally well-satisfied by the obvious improvement of symptoms.

Discussion

Constipation is a common abdominal symptom in childhood [1, 2]. Pediatric surgeons are frequently consulted about children with constipation to search for potentially correctable surgical causes. Suspected Hirschsprung disease is among the common causes of referral. Cases with Hirschsprung disease have a normal anal canal but with defective intestinal motility affecting essentially the rectum and extending to a variable distance along the rest of the intestine [1]. The perineum appears normal, while the diagnosis depends on typical history, characteristic contrast enema findings, and histopathological evidence of absent ganglion cells in the affected segment. A successful pull-through procedure eliminating the aganglionic segment can offer a permanent cure for constipation in Hirschsprung patients [1]. Congenital anal stenosis is another correctable surgical cause for constipation; the diagnosis is essentially clinical based on the obvious abnormal shape and location of the anus [8].

Constipation caused by the anterior location of the anus without stenosis has been reported by Hendren in a large number of children [3], and also by Leape and Ramenofsky in the same year 1978 [4]. Clinical presentation was very characteristic and included constipation dating since early infancy associated with severe straining and pain at defecation. Posterior anoplasty (± sphincterotomy) resulted in immediate relief of symptoms [3, 4]. The diagnosis was frequently overlooked (unrecognized) due to a lack of stenosis and subtle deviation of the anal position from the norm that may be considered a sort of normal individual variation [3]. Hendren described the condition as 'the mild end of the imperforate anus spectrum' and recommended posterior anoplasty to relieve constipation after the exclusion of Hirschsprung disease [3].

Pena firmly denied the term 'anterior ectopic anus' when he proposed a unified term "rectoperineal fistula' that would stand for most of anorectal anomalies at the mild end of the spectrum [9]. Later, in 2005, the pediatric surgical gathering at Krickenbeck adopted Dr. Pena's recommendations; and 'perineal fistula' became one of the major types of anorectal anomalies in both males and females [10]. It may be inconvenient to include all cases described in this report (as well as those in previous ones [3–5]) under the category of 'perineal fistula'. The latter is usually obviously displaced in the perineum at least partially outside the borders of the sphincter and often has a narrow caliber. That was Dr. Pena's argument for calling it a fistula rather than an anus; the latter should be of normal caliber and surrounded by sphincters [9].

The process of defecation is controlled by a complex structural arrangement of muscles and ligaments around the anorectum [11]. In a previous study, we discussed different arguments and theories about obstructed defecation with anorectal anomalies [7]. Similarly, Upadhyaya tried to explain the cause of constipation in children with mild anterior anal misplacement that fail to evacuate their bowel despite severe straining and without apparent cause [5]. He suggested that the primary defect was involving abnormal disposition of the anal sphincters around the mid-anal canal. This segment turns out to be incapable of active propulsion and obstructing the process of defecation [5]. Posterior anoplasty and sphincterotomy remove the lower loop of the external sphincter and allow direct expulsion of feces from the distended mid-anal canal [5]. Interestingly, our observations in studying the pelvic MRI of these patients were consistent with the interpretations of Upadhyaya who suggested the primary defect to be in the mid-anal canal [5]. Despite the subtle external deviation in the position of the anus in these children, the anatomical deviation was more pronounced at deeper levels. In the axial planes, the deeper anal canal was seen anteriorly displaced in relation to the transverse perineal muscles and ischial tuberosities (Figs. 5 and 6). Moreover, in the mid-sagittal plane (Figs. 4 and 5), the length of the anal canal was obviously elongated by the descent of the anus away from the lower end of the pubic symphysis in comparison to normal control subjects of similar age.

We do agree with Dr. Hendren remarks [3] that such cases (which are described in this report) represent the mild end of the spectrum of anorectal anomalies. Although the mechanism of corrective surgery may not be fully understood [5, 7], most cases responded well to posterior/cutback anoplasty through the external anal sphincter (posterior external sphincterotomy) [3–5]. This condition should be distinguished from another different category of patients with intractable constipation due to internal anal sphincter achalasia [12]. The latter group has a different clinical picture (normally located anus) and can be diagnosed by anal manometry. Also, they have different imaging findings: thickening of their internal anal sphincter via ultrasound or MRI [2, 13, 14]. The treatment is different, which includes either internal sphincter myectomy above the dentate line via trans-anal approach or botox injection [15].

The study is limited by its retrospective nature and limited number of cases available at long-term follow-up. Also, there may be no consensus on the diagnosis based on the external appearance alone making it one of the

most difficult cases to manage when we cannot exclude the risk of selection bias. However, suffering at defecation is strikingly obvious in such a group of patients, and is well documented in previous reports as well [3–5]. Posterior anoplasty has stood the test time as an effective, safe, and simple procedure to treat such conditions although the exact pathogenesis remains not fully understood [3–5]. Fortunately, in our report, MRI examinations have unveiled pelvic anatomical alterations at deeper levels that can add further confirmation/evidence to the presence of a structural abnormality. More sophisticated functional studies (manometric, dynamic MRI/defecography) may help to better understand the pathogenesis of constipation which will need older and cooperative children.

Conclusion

Mild unrecognized forms of anorectal anomalies may present with idiopathic constipation. Severe straining at defecation is an important clue. The subtle abnormality on perineal examination can be supported by pelvic MRI unveiling more structural abnormalities at a deeper level. Posterior anoplasty offers relief of symptoms in most cases.

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Authors' contributions
Data acquisition, analysis, and interpretation were performed by all authors (AA; AA; OH; SM). AA made the drafting of the manuscript. All authors have read and approved the final manuscript.

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Availability of data and materials
The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate
All methods were carried out in accordance with relevant guidelines and regulations. An informed parental consent was taken before operation in all cases. Owing to the retrospective nature of the study, an IRB number was not required, and the study was considered exempt research by the scientific/ethical committee of the Pediatric Surgery department (Faculty of Medicine; Ain-Shams University).

Consent for publication
Not applicable. Patient identity did not appear in any part of the manuscript; therefore, consent for publication was not required.

Competing interests
The authors read and approved the final manuscript.

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