Archivos de Medicina
Reporte de Caso

“Congenital rectal atresia: a rare malformation that may go unnoticed. Case report

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Received for publication: 28 June 2025. Revised version: 15 November 2025. Approved for publication: 16 April 2026.

Modelo de citación:

Oliveros-Rivero J., Fajardo-Ponce G. Congenital rectal atresia: a rare malformation that may go unnoticed. Case report. Arch Med (Manizales). 2026;26(1). https://doi.org/10.30554/archmed.26.1.5433.2026

Abstract

Introduction: *Congenital rectal atresia is a rare anorectal malformation, characterised by a normal-appearing anus with interruption of the rectal canal a few centimetres from the dentate line. Its clinical signs mimic other causes of neonatal intestinal obstruction, and diagnosis may be delayed if anal patency is not properly assessed.* **Case report:** *A full-term male newborn presenting with abdominal distension, vomiting and absence of meconium passage. The initial assessment reported a “permeable anus”, leading to a misdiagnosis of necrotising enterocolitis. At 3 days of age, a laparotomy was performed, revealing a colonic perforation. A second operation confirmed the diagnosis of rectal atresia. Surgical management was carried out in stages: colostomy, posterior sagittal anorectoplasty with end-to-end anastomosis, and closure of the colostomy at 11 months. The patient had a favourable clinical and functional outcome.* **Discussion:** *The normal appearance of the anus can delay diagnosis. The neonatal physical examination should include instrumental verification of anal patency. The staged surgical approach allowed for the patient’s stabilisation, definitive diagnosis and successful reconstruction, in line with reports in the literature.* **Conclusion:** *Rectal atresia should be suspected in neonates with signs of lower intestinal obstruction despite a normal-appearing anus. Early diagnosis and timely surgical management are essential for a favourable outcome.*

Keywords: *Congenital rectal atresia; Anorectal malformations; Neonatal intestinal obstruction; Late diagnosis; Paediatric surgery.*

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Atresia rectal congénita: una malformación rara que puede pasar desapercibida

Resumen

Introducción: *La atresia rectal congénita es una malformación anorrectal rara, caracterizada por un ano de aspecto normal con interrupción del canal rectal a pocos centímetros de la línea dentada. Sus signos clínicos simulan otras causas de obstrucción intestinal neonatal y el diagnóstico puede retrasarse si no se evalúa adecuadamente la permeabilidad anal.* **Presentación del caso:** *Recién nacido a término, masculino, con distensión abdominal, vómitos y ausencia de eliminación de meconio. La evaluación inicial reportó un "ano permeable", lo que condujo a un diagnóstico erróneo de enterocolitis necrotizante. A los 3 días de vida se realizó laparotomía con hallazgo de perforación colónica. Una segunda intervención confirmó el diagnóstico de atresia rectal. El manejo quirúrgico se efectuó por etapas: colostomía, anorrectoplastia sagital posterior con anastomosis término-terminal y cierre de colostomía a los 11 meses. El paciente presentó evolución clínica y funcional favorable.* **Discusión:** *La apariencia normal del ano puede retrasar el diagnóstico. El examen físico neonatal debe incluir la verificación instrumental de la permeabilidad anal. El abordaje quirúrgico escalonado permitió la estabilización del paciente, el diagnóstico definitivo y la reconstrucción exitosa, en concordancia con lo reportado en la literatura.* **Conclusión:** *Debe sospecharse atresia rectal en neonatos con signos de obstrucción intestinal baja a pesar de un ano de apariencia normal. El diagnóstico precoz y el manejo quirúrgico oportuno son fundamentales para un desenlace favorable.*

Palabras clave: *Atresia rectal congénita; Malformaciones anorrectales; Obstrucción intestinal neonatal; Diagnóstico tardío; Cirugía pediátrica.*

Atresia retal congénita: uma malformação rara que pode passar despercebida

Resumo

Introdução: *A atresia retal congénita é uma malformação anorretal rara, caracterizada por um ânus de aspeto normal com interrupção do canal retal a poucos centímetros da linha dentada. Os seus sinais clínicos assemelham-se a outras causas de obstrução intestinal neonatal e o diagnóstico pode ser atrasado se a permeabilidade anal não for avaliada adequadamente.* **Apresentação do caso:** *Recém-nascido a termo, do sexo masculino, com distensão abdominal, vómitos e ausência de eliminação de mecónio. A avaliação inicial indicou um «ânus permeável», o que levou a um diagnóstico errado de enterocolite necrosante. Aos 3 dias de vida, foi realizada uma laparotomia com descoberta de perfuração colónica. Uma segunda intervenção confirmou o diagnóstico de atresia retal. O tratamento cirúrgico foi realizado por etapas: colostomia, anorectoplastia sagital posterior com anastomose término-terminal e fechamento da colostomia aos 11 meses. O paciente apresentou evolução clínica e funcional favorável.* **Discussão:** *A aparência normal do ânus pode atrasar*

o diagnóstico. O exame físico neonatal deve incluir a verificação instrumental da permeabilidade anal. A abordagem cirúrgica escalonada permitiu a estabilização do paciente, o diagnóstico definitivo e a reconstrução bem-sucedida, em concordância com o relatado na literatura. **Conclusão:** *Deve-se suspeitar de atresia retal em recém-nascidos com sinais de obstrução intestinal baixa, apesar de um ânus de aparência normal. O diagnóstico precoce e o tratamento cirúrgico oportuno são fundamentais para um desfecho favorável.*

Palavras-chave: *Atresia retal congénita; Malformações anorretais; Obstrução intestinal neonatal; Diagnóstico tardio; Cirurgia pediátrica.*

Introduction

congenital rectal atresia is a rare anorectal malformation, accounting for between 0.3% and 2% of such defects [1,2]. It is characterised by a normal-appearing anus and an interruption of the rectal canal a few centimetres from the dentate line [2]. This anatomical appearance can delay diagnosis if a thorough perineal examination is not performed, thereby increasing the risk of complications such as intestinal perforation [1,3,4].

The clinical presentation includes abdominal distension, vomiting and failure to pass meconium, symptoms common to other causes of lower intestinal obstruction [3,4]. The diagnosis is confirmed by digital rectal examination, radiological investigations and contrast studies. Although numerous classifications have been proposed, there is still no standardised system [2].

Surgical treatment is tailored to the individual and includes techniques such as posterior sagittal rectoanal anastomosis, transanal or combined approaches, with good functional outcomes, although constipation is common during follow-up [5–7].

This report describes a case of congenital rectal atresia with a delayed diagnosis, highlighting the importance of neonatal clinical assessment and a staged surgical approach.

Case presentation

a male newborn, the sole product of a 39-week pregnancy delivered by caesarean section, with an Apgar score of 8-9-9, weighing 3180 g and with no relevant maternal or perinatal history. At , the initial physical assessment reported a “permeable anus”; however, he did not pass meconium within the first 24 hours of life.

At 30 hours of life, he developed vomiting with streaks of blood, progressive abdominal distension and chocolate-coloured gastric output via an orogastric tube, leading to a suspicion of necrotising enterocolitis and the initiation of medical treatment.

In view of the progressive clinical deterioration, with increased gastric output, enlargement of the abdominal circumference and radiological findings of dilated intestinal loops with oedema and air-fluid levels (Figure 1), a diagnosis of intestinal obstruction was considered and he underwent surgery at 3 days of life. During exploratory laparotomy, a 5 cm colonic perforation was identified in the middle portion of the transverse colon, involving more than 80% of its circumference, with leakage of intestinal contents. A colo-colonic anastomosis was performed using 4-0 Vicryl sutures, and a Penrose drain was placed. The cause of the distal obstruction was not identified during this procedure.

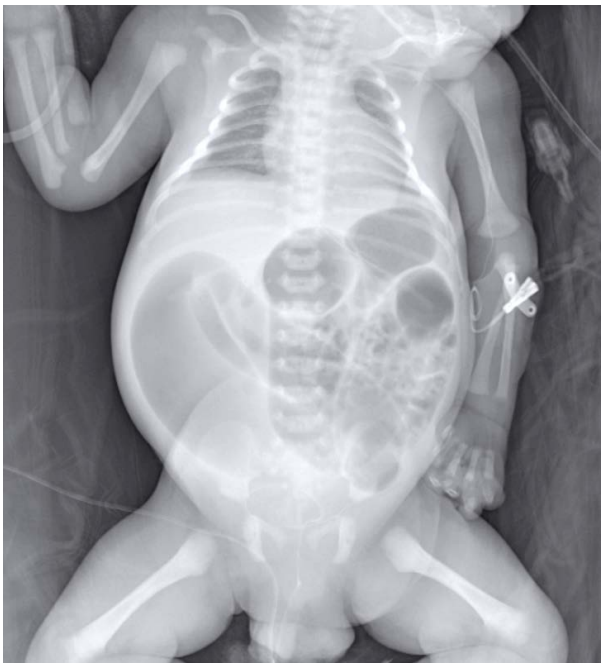

Figure 1. Preoperative X-ray

Five days later, due to persistent abdominal distension and dark bilious discharge via an orogastric tube, he underwent a second operation. During the second laparotomy, dehiscence of the previous anastomosis and dilation of the descending colon containing intestinal contents were observed. As the diagnosis of the malformation had not been established initially and there was a previous colonic perforation, it was decided to perform a double-outlet diverting colostomy, utilising the previous surgical site. The proximal stump (ascending colon) and the distal stump (descending colon) were brought out on opposite flanks, allowing for effective diversion without the need for further resection or anastomosis. Although this type of procedure does not correspond to the usual location in the lower left quadrant, it was considered the safest option given the local conditions. The distal segment was irrigated with saline solution, with no discharge through the anal canal, confirming the diagnosis of previously unrecognised rectal atresia. The procedure was completed with peritoneal lavage. The patient was transferred to the neonatal intensive care unit, where he remained intubated and on mechanical ventilation for three days.

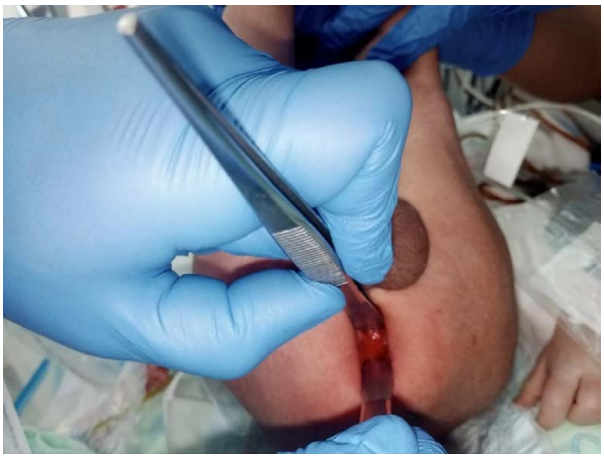

Figure 2. Rectal atresia on physical examination

The colostomy was functional 12 hours after the end of surgery. On the third postoperative day, the patient was extubated and enteral feeding was restarted with good tolerance. He completed a course of antibiotics with piperacillin/tazobactam and metronidazole. During hospitalisation, additional imaging studies were performed to rule out associated malformations; for instance, spinal canal ultrasound showed no abnormalities suggestive of tethered cord, and upper and lower abdominal ultrasound revealed kidneys with normal morphology and echogenicity, without urinary tract dilation or presacral masses. On the fifth postoperative day, the patient was discharged due to an appropriate clinical course.

At 6 months of age, a contrast study via a distal colostomy was performed, ruling out fistulas into the urinary tract and confirming rectal atresia (Figure 3). Subsequently, a recto-anal anastomosis was performed via a posterior sagittal approach with preservation of the anal canal (Figure 4), following the technique described by Peña et al. [8], which allows for complete exposure of the sphincteric complex and precise anatomical reconstruction of the recto-anal canal. During the procedure, a type I rectal atresia was identified according to the classification by

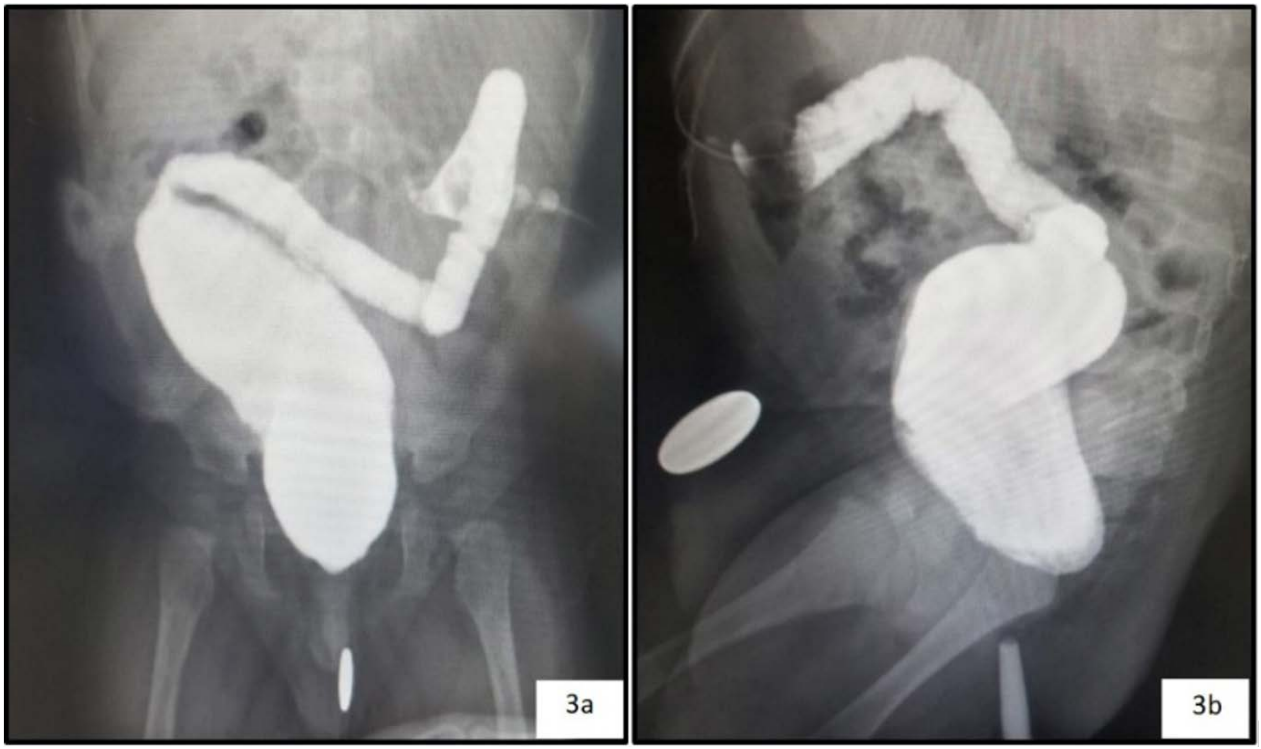

Figure: 3a. AP view of the distal cologram; **3b.** Lateral view of the distal cologram.

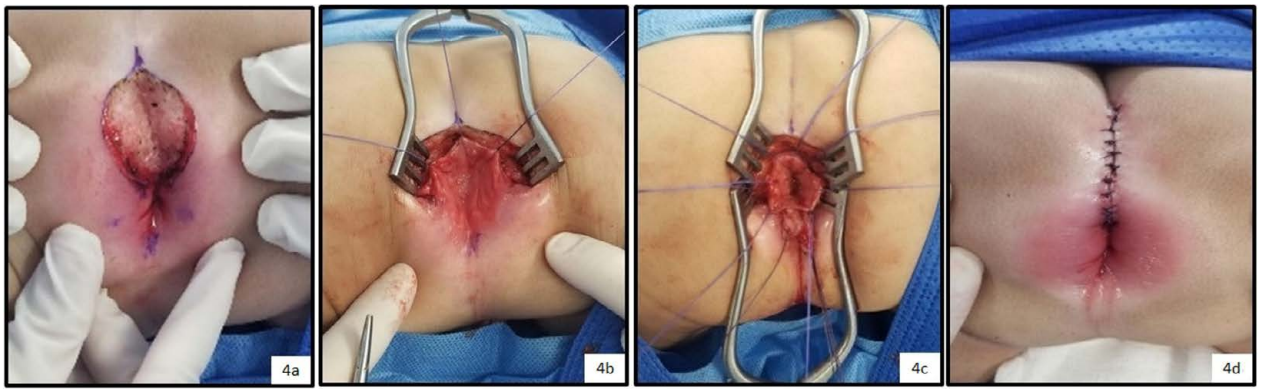

Figure: 4a. PSAP incision; **4b.** Rectal atresia; **4c.** opening of the rectum and start of the end-to-end anastomosis; **4d.** surgery completed

Peña and De Vries (1982), characterised by the presence of a fibrous septum approximately 1 cm in length separating the proximal and distal segments, both of similar calibre and with adequate blood supply. This finding corresponds to the septal or membranous variant, which allowed a direct end-to-end anastomosis to be performed without the need for intestinal tapering. A biopsy of the distal colon was taken for histopathological examination, which revealed the presence of normal ganglion cells, ruling out Hirschsprung's disease. The patient began feeding 12 hours postoperatively, completed a course of antibiotics with ceftriaxone and amikacin, and was discharged on the seventh day. Outpatient follow-up included the use of Hegar dilators for anal dilation according to the age-specific protocol.

Finally, after completing the course of anal dilations with Hegar dilators as recommended in the literature [9] and confirming the absence of stenosis by digital rectal examination, it was decided not to perform the distal cologram. At 11 months of age, an exploratory laparotomy was performed to close the colostomy, with a latero-lateral anastomosis of the transverse colon using a mechanical stapler and placement of a Blake drain. The patient resumed oral intake on the fourth postoperative day and had bowel movements via the anus on the fifth day. The drain was removed on the seventh day, and the patient was discharged on the eighth day with outpatient follow-up at 1, 3 and 6 months, showing good clinical and functional progress.

Discussion

this case demonstrates how an incomplete initial clinical assessment can lead to diagnostic delays in rare neonatal conditions such as rectal atresia. Despite the presence of classic symptoms such as abdominal distension, vomiting and failure to pass meconium, the finding of a normal-appearing anus led to a misdiagnosis of necrotising enterocolitis. The case reaffirms that the neonatal physical examination should include not only perineal inspection, but also verification of anal patency using appropriate instruments [1–3,10].

Most cases of rectal atresia in the literature present in the neonatal period, with difficulty passing a thermometer or dilator beyond 1.5–2 cm from the anal margin [2,6,11]. As in our patient, the normal appearance of the anus delays diagnosis until complications such as colonic perforation become evident or a lack of clinical improvement with medical management is observed. Furthermore, it has been reported that a diversion colostomy not only allows for decompression but also facilitates contrast studies that confirm the absence of fistulas [6,7].

Regarding the surgical approach, multiple techniques have been described: posterior sagittal anorectoplasty, transanal anastomosis, laparoscopically assisted anastomosis, and other less common techniques such as magnetic compression anastomosis [5,6,12,13]. Although there is no universal consensus, the primary objective is to preserve the sphincter apparatus and achieve functional continence. In our experience, the staged technique allowed for stabilisation, accurate diagnosis, and reconstruction with good clinical and functional outcomes, which is consistent with reports in the literature [5,6,11].

This clinical case contributes to the literature by highlighting the importance of timely diagnosis of this rare condition, as well as the value of a staged surgical approach in experienced centres. Correct identification of cases with a normal anus should lead to the exclusion of rectal atresia when there is low-level obstruction, and paediatric surgeons should be familiar with the various repair techniques and their selection based on the anatomical characteristics of each patient.

A comparison of our case with previously published reports is summarised in Table 1.

Conclusion

congenital rectal atresia is a rare malformation that can go undetected in the presence of a normal-appearing anus. Early diagnosis through careful physical examination and a staged surgical approach can prevent complications and lead to good functional outcomes.

CONFLICTS OF INTEREST: The authors declare that they have no conflicts of interest.

FUNDING SOURCES: None.

Table 1. Comparative review of clinical cases of congenital rectal atresia

No.	Author / Year	Age at the time of surgery	Sex	Weight at the time of surgery	Previous colostomy	Surgical technique	Outcome
1	Laamrani et al, 20143	20 days	F	-	No	ATT- ARSP	Good
2	Hamid et al, 20224	3 months	M	2.5 kg	Yes	ATT- ARSP	Good
		3 months	M	3 kg	Yes	ATT- ARSP	Good
		1 day	F	3 kg	Yes	ATT- ARSP	Good
3	from Beaufort et al, 20235	70 patients (1 day to 6 years of age)	M (n=37) F(n=21) N/A (n=12)	N/E	Yes (n=56) No (n=14)	ATT- ARSP (n=43) DTA (n=18) Local resection (n=8) Magnamosis (n=1)	Good (n=66) Poor (n=4)
4	Shehata et al, 20236	6 months	M	-	Yes	ART	Good
		6 months	M	-	Yes	ART	Good
		6 months	M	-	Yes	ART	Good
		6 months	F	-	Yes	ART	Good
5	by Beaufort et al, 20237	18 patients 2–6 months (median 5 months)	M (n=10) F(n=8)	N/A	Yes (n=16) No (n=2)	ATT- ARSP (n=9) DTA (n=1) ART (n=1) ACR (n=1) ATA + D (n=1) N/A (n=5)	Good (n=11) Poor (n=2) Died before surgery (n=1) N/A (n=4)
6	Sharma et al, 20178	1 day	N/A	-	Yes	DAP	Good
		1 day	N/A	-	Yes	DAP	Good
		1 day	N/A	-	Yes	ATT- ARSP	Good
		1 day	N/A	-	Yes	ATT- ARSP	Good
		1 day	N/A	-	Yes	ATO - ARSP	Good
7	Gieballa et al, 20189	7 months	M	-	Yes	DTA	Good
		6 months	M	-	Yes	DTA	Good
		10 months	M	-	Yes	DTA	Good
8	Jiang et al, 202410	5 months	F	-	Yes	DTA	Good
9	Mehmetoğlu et al, 201811	9 months	M	-	Yes	ATT- ARSP	Good
10	Dias et al, 198214	12 months	M	-	Yes	ATT-ASU	Good
11	Upadhyaya, 199015	42 months	M	-	Yes	ART	Good
		6 months	M	-	Yes	ART	Good
12	Kisra et al, 200516	3 months	M	-	Yes	ATT- ARSP	Mild stenosis resolved with dilation
		3 months	M	-	Yes	ATT- ARSP	Mild stenosis resolved with dilation
		3 months	M	-	Yes	ATT- ARSP	Good
		3 months	M	-	Yes	ATT- ARSP	Good
13	Nguyen TL et al, 200717	3 months	F	2.3 kg	Yes	ART-LA	Good
		3 months	F	2.7 kg	Yes	ART-LA	Good
14	Hamrick et al, 201218	17 patients (aged 2 days to 6 years)	M(n=10) F(n=7)	-	Yes (n=13) No (n=4)	ATT- ARSP (n=4) ATO- ARSP (n=13)	Good, 5 patients (29%) with mild constipation
15	Our case	4 months	M	5.1 kg	Yes	ATT- ARSP	Good

ATT-ARSP: End-to-end anastomosis following posterior sagittal anorectoplasty; ART: Transanal rectoanal anastomosis; DAP: Abdominoperineal approach; N/E: Not specified; DTA: Transanal endorectal approach; ACR: Colorectal anastomosis; ATA + D: Transanal opening under anaesthesia + dilation; ATO-ARSP: End-to-oblique anastomosis following posterior sagittal anorectoplasty; ATT-ASU (End-to-end anastomosis via single sacral approach); ART-LA: Laparoscopically assisted transanal rectoanal anastomosis.

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