



## Neonatal colon perforation due to anorectal malformations: Can it be avoided?

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**Core tip:** Anorectal malformations (ARM) are common anomalies observed in neonates. The delay in diagnosing a neonate with ARM results in significant complications, occasionally life-threatening morbidity, such as colon perforations. However, delayed diagnosis of ARM seems not the unique factor leading to colonic perforation, deficiency of musculature in the gut wall may also contribute. Colonic perforation due to ARM may not be avoided completely; however, early diagnosis is essential in assuring better outcomes with surgical management.

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### Abstract

Anorectal malformations (ARM) are common anomalies in neonates. Diagnostic and therapeutic delays in the management of ARM may lead to colonic perforation, and even death. Physical examination of the perineum is often sufficient to diagnose ARM in neonates. Notwithstanding, delayed diagnosis of ARM has become increasingly familiar to surgeons, as evidenced by the number of recent publications on this topic in the literature. In this commentary, we discuss spontaneous colonic perforation due to delayed diagnosis of ARM in neonates, and highlight the importance of early diagnosis in assuring good outcomes with surgical management. At this point, a thorough examination of the perineum during the initial newborn assessment is mandatory, particularly in those patients presenting with abdominal signs or symptoms.

### COMMENTARY ON HOT TOPICS

We have read with great interest the recent article by Kapadnis *et al* describing a 2.5 kg neonate presenting after 72 h with sigmoid colon perforation due to anorectal malformation (ARM). Delayed diagnosis of ARM has become increasingly familiar to surgeons, as evidenced by the number of recent publications on this topic in the literature<sup>[1,2]</sup>. Despite the recommendations for peri-natal assessment<sup>[3]</sup>, the overall incidence of a delayed diagnosis has recently been reported to be as high as 21.2%<sup>[2]</sup>. The delay in diagnosing a neonate with ARM results in significant complications, occasionally life-threatening morbidity, such as colon perforations. Spontaneous perforation of the colon is estimated to occur in 2% of neonates

**Table 1** Classification of intestinal perforations complicated with anorectal malformations (*n* = 25)

Type of perforation	Frequency <sup>1</sup>	Description	Recommended management
Type 1		Perforation occurring before relief of obstruction	
Type 1a	16%	Involving cecum or proximal ascending colon	Cecostomy + distal colostomy
Type 1b	8%	Involving transverse colon including the 2 flexures	Exteriorization of perforation (as colostomy)
Type 1c	60%	Involving distal sigmoid or rectum	Closure of perforation + proximal colostomy
Type 1d	4%	Other sites such as vagina in cloaca	Closure of perforation + proximal colostomy
Type 2	12%	Perforation occurring in the postoperative period	Exteriorization of the perforation site

<sup>1</sup>Calculated by combining the 17 cases reported in the literature and the authors' series.

with ARM, and the incidence rises to 9.5% when the diagnosis is delayed<sup>[2]</sup>. Thus, it seems crucial to diagnose and treat ARM early to avoid colon perforation.

ARMs are common anomalies observed in neonates<sup>[4]</sup>. The reported incidence ranges between 1:3300 and 1:5000 live births. In Western countries, there is a male preponderance with 55%-70% of the patients in larger series being males<sup>[6]</sup>. They vary in severity from mild anal stenosis to complete caudal regression. These disorders usually require surgical intervention in the neonatal period and postoperative follow-up to obtain and maintain fecal and urinary continence. Diagnostic and therapeutic delays in the management of ARM may lead to complications such as sepsis, aspiration, abdominal distension, colonic perforation, respiratory embarrassment, electrolyte imbalance, and even death. The diagnosis of ARM is usually made at birth or shortly thereafter physical examination. Standardized national and international guidelines recommend a routine physical examination of all newborns within the first 48 h of life<sup>[3,5]</sup>. It has been reported that the median age at diagnosis of perforation in ARM cases was 48 h<sup>[6]</sup>. Generally, delayed diagnosis of ARM is defined as a diagnosis made after the first 48 h<sup>[2]</sup>. Undoubtedly, the necessity to diagnose ARM in a timely manner is reliant on a comprehensive neonatal examination performed by a pediatrician or pediatric trainee with sufficient experience. Furthermore, neonatal examination of all newborns should be made within the first 48 h of life. Increasing the awareness among pediatricians of the challenges and complications due to delayed ARM diagnosis may be the important first step. Additional training to adequately diagnose ARM, or change current guidelines to explicitly rule out ARM is also required. Some researchers believe that a higher incidence of associated anomalies may promote earlier diagnosis of the ARM<sup>[2]</sup>, whereas others failed to confirm this hypothesis<sup>[7]</sup>. Wilson *et al*<sup>[7]</sup> believed that the only significant predictor of delayed diagnosis of ARM was a failure to receive a comprehensive neonatal examination within 48 h, reiterating that timely diagnosis of ARM is best achieved by adequate clinical examination.

However, colonic perforations cannot be simply attributed to the delayed diagnosis or treatment of ARM, because there are a few case reports of bowel rupture occurring during intrauterine life<sup>[8]</sup>. Based on their research and review of the literature, Raveenthiran<sup>[6]</sup> summarized

two distinct patterns of perforations involving four different sites and recommended management (Table 1). Approximately 88% of perforations are of type 1, whereas only 12% are of type 2. Among the type 1 perforations, 60% occur in the rectum and sigmoid colon<sup>[6]</sup>. This difference suggests that the mechanism of perforation could be different for the two types. A higher ratio of rectosigmoid perforation in ARM implies an embryologic origin. As ARM is a developmental field defect, the tail end of the gut can be expected to have deficiency of musculature. The downstream obstruction leads to increased intraluminal pressure, and this, along with the muscular deficiency, is probably responsible for more frequent rupture of the rectum in ARM. Mathur *et al*<sup>[9]</sup> reported five perforations (6.5%) among 77 cases of ARM with congenital pouch colon (CPC). A high incidence of bowel perforation in CPC also favors the muscular deficiency theory. At this point, delayed diagnosis of ARM seems not the unique factor leading to colonic perforation.

Despite the fact that not all colonic perforations are the result of delayed diagnosis of ARM, the majority are, and early diagnosis is essential so that surgical management can commence to achieve better outcomes. At this point, a thorough examination of the perineum during the initial newborn assessment is mandatory, particularly in those patients presenting with abdominal signs or symptoms.

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