Intestinal perforation secondary to congenital segmental agenesis of intestinal muscularis

Ahmed H. Al-Salem, FRCSI, Shahzad S. Qureshi, M. MED.

Intestinal muscularis is a rare disorder of unknown etiology. It is a rare cause of perforation in newborns which may resemble necrotizing enterocolitis (NEC). The diagnosis is by segmental absence of the muscularis propria, normal intact mucosa, submucosa and serosa.

ABSTRACT

Congenital segmental abscess with intestinal obstruction or perforation can be confirmed histologically by the absence of muscularis.
literature.\textsuperscript{5,6,8,9,10} Classically the disease is segmental and has been reported to affect the stomach,\textsuperscript{3,11,12} small,\textsuperscript{5,6,8,10} and large intestine.\textsuperscript{13} Although congenital segmental absence of the muscular layer of the intestine can affect any part from the stomach to the colon, it is most commonly seen affecting the small intestine and the distal ileum in particular.\textsuperscript{5,8,10} In reviewing the literature, 38 cases were found in the English literature,\textsuperscript{7,11,12} 2 in the colon\textsuperscript{13} and 29 in the small intestine.\textsuperscript{5,6,8,10,14} Classically it presents as intestinal perforation or obstruction with or without intussusception. The age at presentation ranged from 1 day to 14 years\textsuperscript{6} but the majority present in the neonatal period and most of them are prematures.\textsuperscript{5} In two patients, the defect was found incidentally during repair of an incarcerated inguinal hernia.\textsuperscript{15} When it presents with perforation in the newborn period it is usually confused with necrotizing enterocolitis and should be considered in the differential diagnosis of neonatal intestinal obstruction. Intraoperatively, the bowel is frequently described as remarkably transparent as in our case.\textsuperscript{7,10,15}

The diagnosis is confirmed histologically. The bowel will show normal intact mucosa with preservation of the submucosal Meissner's plexus, complete absence of the muscularis propria and normal serosa. Humphry et al\textsuperscript{16} reported a 14-month old girl with absence of only the circular muscle of the small bowel, and McCarthy et al\textsuperscript{10} reported a 4 day old newborn who had absence of muscularis mucosa as well. Except for these two reports, all reported cases showed complete absence of the muscularis propria only. The absence of inflammation, hemorrhage and necrosis are important histological findings that differentiate this from necrotizing enterocolitis.\textsuperscript{6}

The exact etiology of this condition is not known. It is presumed by many to be congenital in origin. Carrol, Jr\textsuperscript{17} suggested ischemia to be the etiologic factor rather than a congenital absence of the musculature. Alvarez et al\textsuperscript{14} in describing a case of small intestinal atresia and segmental absence of muscular coats suggested in utero vascular accident as a possible etiology for both conditions. Although ischemia is considered by some authors as the explanation for this, we agree with Litwin et al\textsuperscript{9} that it is difficult to explain the muscular damage on this basis while the mucosa which is more sensitive to ischemia remains intact. Al Awadhi et al\textsuperscript{9} based on the observation that most cases occur in the distal ileum proposed that absence of muscularis propria could be part of the process of regression of the omphalomesenteric remnant leading to the resorption of the muscle of adjacent ileum, but this does not explain its occurrence at other sites of the gastrointestinal tract. The most accepted theory is that based on embryogenesis. In a 7 to 40mm embryos multiple diverticula are present in the intestine especially in the terminal ileum. These diverticula subsequently regress, but if this fails it will lead to failure of condensation of surrounding mesenchyme which usually produce the muscle coats of the bowel leading to focal circumferential absence of the muscularis of the bowel.\textsuperscript{8,10}

Congenital segmental absence of the muscle layer of the intestine is being recognized more frequently nowadays as pediatric surgeons and pathologists are more familiar with the condition and the prognosis has improved markedly over the years.\textsuperscript{5,6,8,10}

References


