Neonatal intestinal perforation secondary to congenital segmental agenesis of intestinal muscularis

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

ABSTRACT
Congenital segmental absence of intestinal musculature is a rare disorder of unknown etiology. It is a rare cause of intestinal obstruction or perforation in newborns which may resemble necrotizing enterocolitis (NEC). The diagnosis is confirmed histologically by segmental absence of the muscularis propria, normal intact mucosa, submucosa and serosa. The absence of inflammation, hemorrhage and necrosis helps to distinguish it from NEC. A case of an infant boy with intestinal perforation secondary to segmental absence of intestinal musculature is presented, the literature is reviewed and possible etiologies are discussed.

Saudi Medical Journal 1997; Vol. 18 (6) : 605-607

Keywords: Absent intestinal muscularis, neonatal gastrointestinal perforation, neonatal intestinal obstruction.

Neonatal focal gastrointestinal perforation is not an uncommon condition which has been described in association with ischemia, Hirschsprung’s disease, indomethacin therapy and gastrointestinal obstruction.1,2,3 Congenital segmental absence of the muscular layer of the small intestine is a very rare cause of intestinal obstruction or perforation in the newborn which is usually confused with neonatal necrotizing enterocolitis.4,5,6 It was first described in 1943 by Herbut.7 Since then 38 cases have been described in the literature. Awareness of this condition is essential for proper diagnosis and management. This is a report of a case of congenital absence of the intestinal muscular layer in a neonate. The literature on the subject is reviewed and possible etiologies are discussed.

Case report. A 30 week gestation boy, one of twins was referred to our hospital at the age of 7 days because of birth asphyxia and respiratory distress. His birth weight was 1.3kg. There was a history of polyhydramnios. He was found to have patent ductus arteriosus. On arrival at our hospital he was ventilated and noted to have abdominal distension. Abdominal x-rays showed pneumoperitoneum. He was diagnosed as a case of necrotizing enterocolitis with perforation. An exploratory laparotomy revealed meconium stained peritoneal fluid. There was an area of perforation in the mid ileum on the antimesenteric border proximal to an area of about 4 cm of transparent bowel. The area of perforation and transparent segment were resected and an end to end anastomosis was carried out. The remaining bowel appeared normal. The histopathology showed complete circumferential absence of muscularis propria (Fig. 1). The serosa, mucosa and submucosa were normal with preservation of the submucosal Meissner’s plexus in the affected area and myenteric plexus as well Meissner’s plexus in the adjacent normal bowel wall. The distal resection margin is unremarkable particularly the muscularis propria which appeared normal (Fig. 2). Post-operatively, the patient continued to require ventilatory support because of birth asphyxia and respiratory distress. He developed sepsis and died on the 19th postoperative day.

Discussion. Congenital segmental absence of the muscular layer of the intestine is a very rare condition.5,6 It was first described in 1943 by Herbut7 who reported a neonate with spontaneous gastric perforation that was devoid of muscles. Since then several case reports appeared in the

From the Departments of Surgery (Al-Salem) and Pathology (Qureshi), Qatif Central Hospital, Qatif.

Received February 1997. Accepted for publication in final form April 1997.

Address correspondence and reprint request to: Dr. Ahmed H. Al-Salem, PO Box 18432, Qatif 31911, Kingdom of Saudi Arabia. Fax No. 03-8553691.
Classically the disease is segmental and has been reported to affect the stomach,\textsuperscript{2,7,11,12} 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, 7 occurred in the stomach,\textsuperscript{2,7,11,12} 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{8} 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{2,7,10,15}

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. Humphery 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 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{8} 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{18} 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 resorb, 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

4. Litwin A, Avidor I, Schujman E. et al. Neonatal intestinal perforation caused by congenital defects of the intestinal