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Congenital absence of intestinal smooth muscle: a case report and review of the literature

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Abstract Herein is reported case of an otherwise healthy full-term infant girl who presented with numerous spontaneous intestinal perforations with congenital absence of intestinal muscularis mucosae and muscularis propria. Few other cases are reported in the English literature with varying presentations. We review those cases, theories of pathogenesis, embryology, and possible connections to various clinical presentations.

1. Case report

A 3-week-old full-term 2.7-kg girl was born without complications by normal spontaneous vaginal delivery to a healthy 21-year-old primagravid mother. She developed watery diarrhea and nonbloody, nonbilious emesis at 2 weeks of life. She was admitted 5 days later for dehydration and was noted to have occult blood-positive stool. She improved clinically with intravenous fluids and antibiotics and tolerated oral feeds. Four days later, she developed significant abdominal distension with pneumatosis intestinalis and pneumoperitoneum on abdominal radiographs. At exploration, significant intraperitoneal air and fluid from a single ascending colon perforation was noted. This was repaired, and a Brooke loop ileostomy was constructed. Four days later, stool was seen in the wound. Reexploration revealed wound dehiscence with additional perforations proximal and distal to the ileostomy. A small segment of ileum was resected, which provided the pathologic findings for this case; a new proximal Brooke ileostomy and distal mucous fistula were created. Five days later, she was again noted to have wound dehiscence, evisceration, and sucus in the abdominal cavity. Reexploration revealed 20 "punched-out" perforations of the small intestines, each located at the junction of the mesentery and small bowel wall. All were oversewn.

Further perforations occurred with the development of multiple enterocutaneous and enteroenteric fistulae, but we did not elect to operate at this time. The patient remained in the pediatric intensive care unit for several months with a course complicated by DIC and TPN cholestasis. Eventually, all distal small bowel, compromising approximately 70% of her small intestine, was resected. She was begun on a small bowel adaptation regimen that was quite successful; her

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proximal small bowel and colon were joined, and she is currently stable and growing nearing 2 years of age.

Histologic examination of the specimen submitted from the first ileal resection showed focal ulceration, disruption of bowel wall, and transmural inflammation consistent with the perforation site. There were 3 different foci of prominent edema of the submucosa with distended focally ectatic vessels (Fig. 1A) and multiple microscopic foci of inflammatory collections (Fig. 1B), composed mainly of small lymphocytes, with accompanying fibroblastic proliferation. The inflammatory foci were scattered and appeared both in submucosal and subserosal distribution, some adjacent to the foci of muscle loss. The myxoinflammatory foci did not invade into the muscle; for example, no definite morphologic evidence of inflammatory myopathy was present. The border of remaining muscle and the lack thereof were very sharp and clean of inflammation. Immunostains for smooth muscle and desmin demonstrated partial or complete loss of the muscle fibers adjacent to the myxoinflammatory pockets as seen in the images (Fig. 1C). Histologic sections show the presence of ganglion cells in both submucosal and myenteric plexuses; some areas of the myenteric (Auerbach) plexus had increased number of ganglion cells in bigger clusters with continuous distribution (based on pure comparative histomorphologic observation; Fig. 1D).

Definitive histologic evidence of findings previously reported in Ehlers-Danlos syndrome (EDS) [1] were not seen, but early manifestations could not be ruled out. Collagen electrophoresis studies showed no abnormalities and eliminated some of the more common forms of EDS. Immunologic evaluation including normal immunoglobulin levels, negative human immunodeficiency virus, and normal complete blood count including differential with adequate lymphocytic response made immunodeficiency syndromes unlikely. No evidence for primary connective tissue disease or vasculitis was discovered.

2. Discussion

Congenital absence of intestinal musculature (CAIM) is a rare but documented entity, although its true incidence is

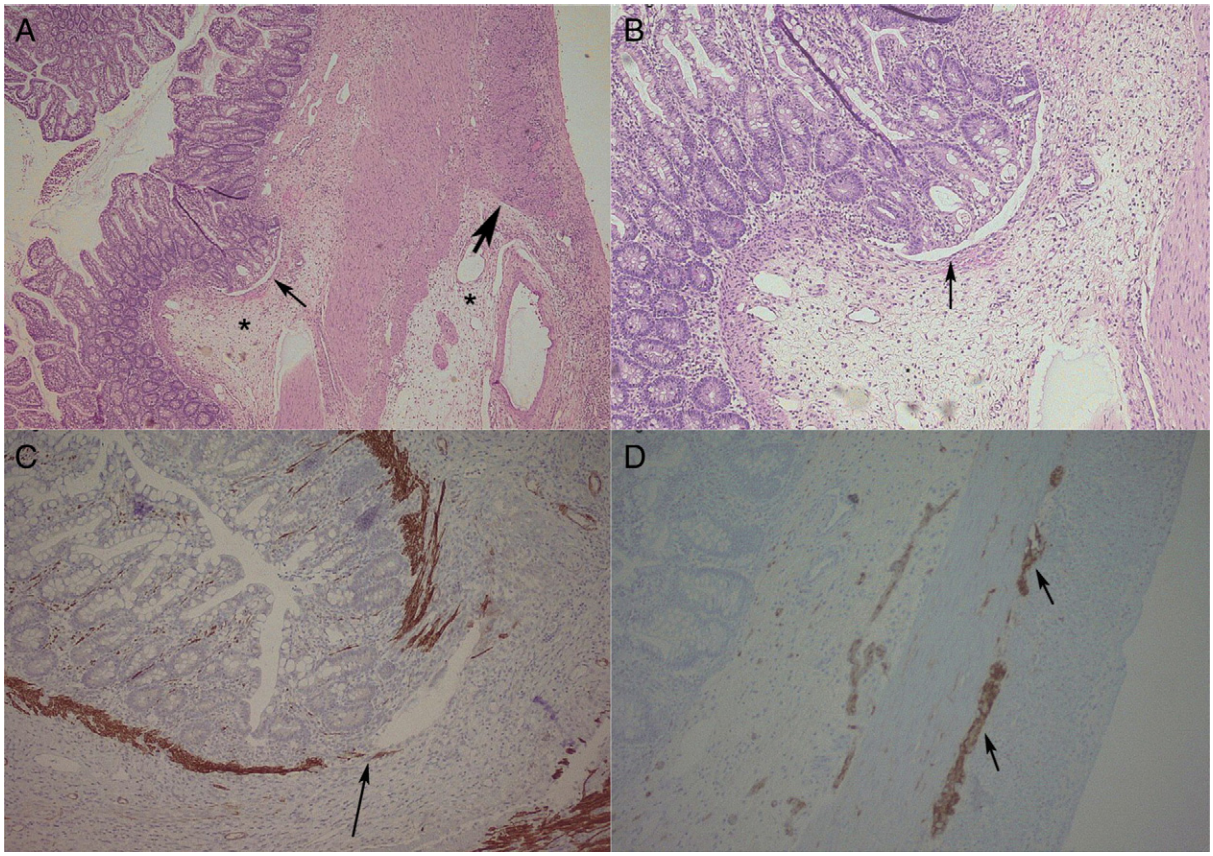

Fig. 1 A, Low power magnification cross section of the small bowel wall, demonstrating intact mucosa with loose edematous foci in submucosal and subserosal location (asterisk). Loss of smooth muscle fibers in both submucosal (thin arrow) and subserosal (thick arrow) distribution is appreciated (original magnification: ×200, H&E stain). B, Higher power of the image in Fig. 1, showing focal complete loss of muscularis mucosae (arrow) and partial prolapse of the mucosa in the loose submucosa (original magnification: ×400, H&E stain). C, Higher power magnification of loss of muscularis mucosae (arrow; original magnification: ×400, desmin immunostain). D, Presence of ganglion cells. Some areas of the myenteric (Auerbach's) plexus had increased number of ganglion cells in bigger clusters with continuous distribution (comparative histomorphologic observation; original magnification: ×400, S-100 immunostain).