LETTER TO THE EDITOR

Segmental Absence of Intestinal Musculature: Unmasking Intestinal Perforation in a Preterm Neonate

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Dear Editor,

We are writing to report a rare case of segmental absence of intestinal musculature (SAIM) diagnosed in a preterm newborn who presented to the emergency department with intestinal perforation. SAIM is a rare congenital anomaly characterized by partial or complete absence of the muscularis propria of the intestinal wall in a segmental pattern.[1] It primarily affects low-birth-weight premature neonates and infants.[1,2] The true incidence of SAIM remains unknown due to its rarity. Clinically, it often presents with spontaneous intestinal perforation (SIP) and closely mimics necrotizing enterocolitis (NEC), making preoperative diagnosis challenging. Given its potential for sudden and severe presentation, SAIM is a critical differential in neonatal gastrointestinal emergencies, necessitating urgent surgical intervention.[3]

A 4-day-old male neonate born preterm at 31 weeks of gestation was referred to emergency care center with progressive abdominal distension and failure to pass meconium since birth. Abdominal radiography revealed pneumoperitoneum, indicating gastrointestinal perforation. An emergency laparotomy was performed, which revealed two jejunal perforations approximately 40 cm and 44.5 cm distal to the duodenojejunal flexure, involving 35 cm of unhealthy segment. Perforation-bearing segment was resected with possible intraoperative suspicion of NEC, followed by anastomosis. Gross examination of the jejunum revealed two perforations of 0.2–0.3 cm diameter along with thinning of the bowel wall. Rest of the specimen showed alternating dilated, thinned segments and relatively normal-caliber segments with intact congested serosal surface (Fig. 1). Microscopy revealed transmural architectural disruption and necrotizing inflammation at perforation site. Adjacent areas showed thinning of the bowel wall with adjacent segments showed multifocal thinning and focal absence of the muscularis propria, predominantly affecting the inner circular layer. The serosa exhibited fibroblastic proliferation, dilated lymphatics, vascular congestion, and marked inflammation in response to the perforation. Ganglion cells were preserved. Segmental pattern of absent musculature was noted throughout the affected resected jejunum (Fig. 2). These findings confirmed the diagnosis of SAIM as the underlying cause of jejunal perforation in this preterm neonate.

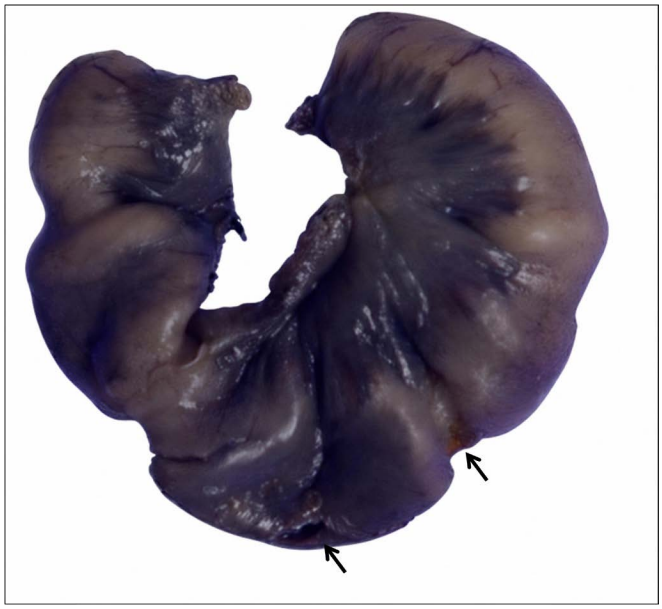

Fig. 1: Specimen of jejunum demonstrating alternate dilated thinned-out and healthy segment along with perforations (arrows)

Postoperatively, the patient resumed bowel movements, tolerated enteral feeds. The initial postoperative course was uneventful, and the patient was discharged 3 weeks after surgery. Unfortunately, the further advanced radioimaging modalities to look for bowel musculature could not be performed due to clinical limitations, as the patient was readmitted to the emergency department with respiratory distress and subsequently succumbed despite medical intervention.

Segmental absence of intestinal musculature is an exceedingly rare but clinically significant congenital anomaly characterized by a localized or segmental deficiency of the muscularis propria of the intestinal wall. The etiology remains poorly understood, with proposed multifactorial mechanisms including defective embryogenesis, intrauterine ischemic events, genetic abnormalities, failed regression of embryonic diverticula, or underdevelopment of muscularis layer.[1,2]

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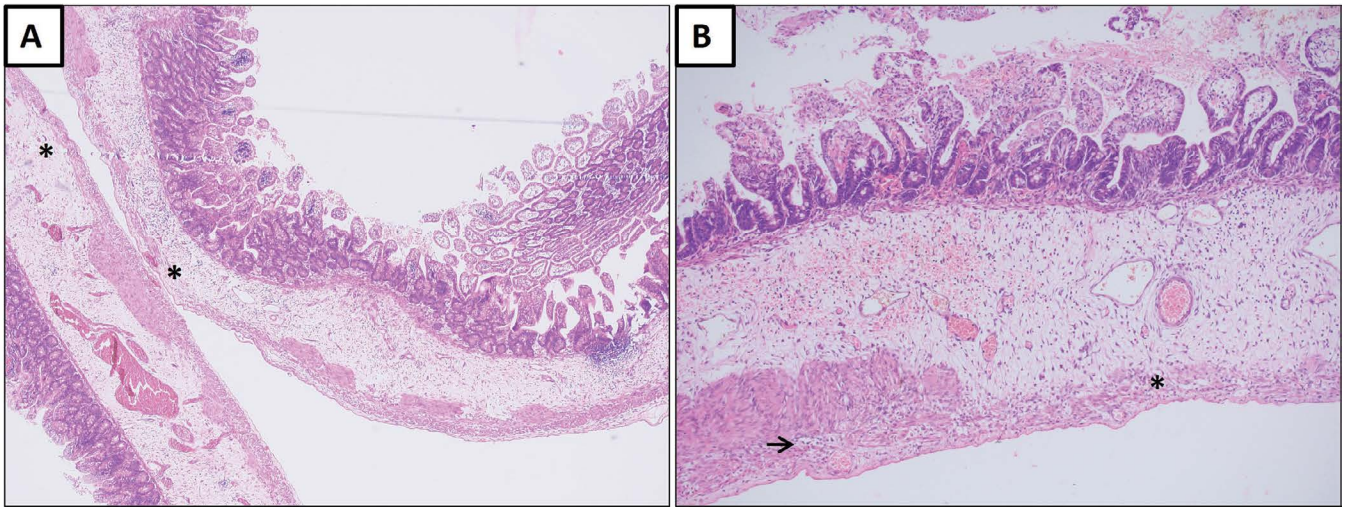

Figs 2A and B: (A) Micrograph from jejunal wall showing intact mucosa and multifocal absence of the muscularis propria and neural plexuses in the thinned segments (asterisks) (H&E, 40×); (B) Higher-power view showing marked submucosal edema, dilated lymphatics, vascular congestion, and preserved ganglion cells within the muscular layer (arrow); the asterisk indicates the area of muscularis propria and neural plexus loss (H&E, 100×)

It primarily affects premature neonates, typically presenting within the first few weeks of life with SIP or, less commonly, intestinal obstruction.[1–4] Although extremely rare, isolated cases have been reported in older children and adults also.[3,5] In a series by Pinzon-Guzman et al., five premature infants presented with intestinal perforation and were subsequently diagnosed with SAIM within the first 2 weeks of life.[1] Similarly, our patient was a preterm neonate who presented with intestinal perforation with peritonitis during the first week of life.

The clinical presentation of SAIM can closely resemble other neonatal abdominal emergencies, particularly NEC, which poses significant diagnostic and management challenges. Shared features such as abdominal distension, feeding intolerance, and radiological evidence of pneumoperitoneum are common to NEC, SIP, and SAIM, making preoperative differentiation difficult.[1,3,6] Notably, SAIM and NEC are not mutually exclusive; cases have been documented where both conditions coexist or where NEC develops following surgical intervention for SAIM-related perforation.[3] In this case, inflammation was confined to the serosal layer of the bowel, likely as a reactive change to the perforation, with no histological features suggestive of NEC observed in the underlying layers.

Owing to its rarity and nonspecific presentation, preoperative diagnosis of SAIM remains exceptionally challenging. Radiological and clinical findings are typically indistinguishable from NEC, SIP, or other causes of neonatal intestinal perforation, leading to potential misdiagnosis.[1–3,7] Although the findings of alternate dilated pattern of unhealthy bowel with multiple perforations, as illustrated in this index case, can raise intraoperative suspicion of this pathology, particularly in preterm neonates, a definitive diagnosis is usually established only postoperatively, based on histopathological evaluation of the resected bowel. In our case, emergency laparotomy was performed for perforation peritonitis without a preoperative etiological diagnosis, and SAIM was confirmed only on microscopic evaluation, underscoring the critical role of pathological assessment in establishing the final diagnosis.

The primary limitation of this report lies in its nature as a single-case study, which inherently restricts the ability to generalize findings or draw causal inferences regarding the pathogenesis, optimal management, or long-term outcomes of SAIM. Additionally, the absence of prenatal or advanced imaging findings specific to SAIM limits insight into potential early diagnostic markers. Genetic or molecular analyses, which might have provided further information regarding underlying etiological mechanisms, were not performed. Furthermore, long-term follow-up was limited due to the unfortunate demise of the patient from nongastrointestinal causes, precluding assessment of late postoperative outcomes.

Delayed or missed recognition of SAIM may result in significant postoperative consequences such as anastomotic leakage, recurrent perforation, or obstruction.[3] This case emphasizes the importance of considering SAIM as a key differential diagnosis in cases of early neonatal intestinal perforation, particularly in preterm infants without classical risk factors for NEC. Increased awareness of this entity may prompt meticulous intraoperative assessment and adequate resection margins, potentially reducing postoperative morbidity.

Optimal management of SAIM requires a multidisciplinary approach, with close collaboration among neonatologists, pediatric surgeons, and pathologists to ensure accurate diagnosis, appropriate treatment, postoperative care, and long-term follow-up. Future studies incorporating multicenter case series, standardized histopathological criteria, and molecular investigations are warranted to improve understanding of the disease spectrum and to explore possibilities for earlier diagnosis.

In conclusion, SAIM is a rare but critical condition affecting particularly preterm neonates. Distinguishing it from NEC or other gastrointestinal emergencies is essential. Awareness of this rare entity is particularly important among pediatric surgeons and pathologists encountering neonatal abdominal emergencies, as early recognition of SAIM has direct implications for surgical decision-making and patient outcomes.

Availability of Data and Materials

There is no data generated during the current study, material prepared is available with the corresponding author.

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Author Contributions

AJ and HJ contributed to the study design and drafting of the manuscript, KU and RPK provided clinical records, and follow-up details of patient and helped in drafting the manuscript, AK, MS, and HJ aided in histopathological data analysis, interpretation, and design of the work by giving valuable insights.

All authors finally approved the version to be published.

AI Use Declaration

No AI-assisted tools were used in the preparation of this manuscript.

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