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Mohamed G El Madawy
Department of General Surgery, Al-Azhar University, New Damietta, Egypt

Corresponding Author:
Mohamed G El Madawy
Department of General Surgery, Al-Azhar University, New Damietta, Egypt

Adult-onset rectal hypoganglionosis: A rare cause of long-standing constipation in a 27-year-old female following appendectomy - a surgical case report

Mohamed G El Madawy

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Abstract
Background: Hypoganglionosis is a rare disorder of enteric innervation characterized by a decreased number of myenteric ganglion cells. It typically presents in childhood, while adult-onset cases are uncommon and often delayed in diagnosis.
Case Presentation: A 27-year-old female, no chronic medical illnesses, presented with progressive, refractory constipation that began after an appendectomy at the age of 18. Over the years, her bowel movements became infrequent until she developed complete stool retention for 40 days prior to presentation, though flatus passage was maintained. Imaging revealed dolichocolon and a short, funnel-shaped distal segment on contrast enema. Anorectal manometry showed increased rectal compliance and paradoxical anal contraction. Histopathology confirmed severe hypoganglionosis involving the rectum and sigmoid colon. Surgical resection with colo-anal anastomosis led to full symptomatic resolution.
Conclusion: Adult-onset rectal hypoganglionosis is rare and frequently misdiagnosed. Full-thickness biopsy remains the diagnostic gold standard. Surgical resection of the affected segment results in excellent functional outcomes when the hypoganglionic segment is completely excised.

Keywords: Adult-onset hypoganglionosis, refractory constipation, dolichocolon, anorectal manometry

Introduction
Hypoganglionosis is an uncommon congenital or acquired abnormality of the enteric nervous system characterized by reduced ganglion cell density in the myenteric plexus, leading to segmental colonic dysmotility and chronic constipation [1]. While congenital forms usually manifest during infancy, adult-onset presentations are rare and can mimic functional bowel disorders or chronic idiopathic constipation [2]. Diagnosis is often delayed because of overlapping symptoms and nonspecific imaging findings. Surgical management remains the definitive treatment when a transitional zone is identified.

Incidence and Prevalence
The true incidence of adult-onset hypoganglionosis remains unknown due to its rarity and diagnostic challenges. Fewer than 100 well-documented cases have been reported worldwide [3], suggesting underdiagnosis in chronic constipation cohorts.

Case Presentation
Patient Profile
A 27-year-old married female, mother of two, with no chronic illnesses, presented with progressive constipation beginning shortly after an appendectomy at age 18. She reported normal bowel habits before surgery.

History
Postoperatively, she developed infrequent bowel motions with continued flatus passage. Symptoms gradually worsened, with intervals of up to one month between bowel movements despite regular use of laxatives and enemas. Previous evaluations (ultrasound, CT, and barium studies) were unremarkable except for dolichocolon. Colonoscopy revealed bilharzial infiltration, treated medically, but symptoms persisted.
Eight years later, she presented with severe constipation (no motion for 40 days), abdominal distension, preserved appetite, and no vomiting.

Physical Examination
Abdomen: distended, tympanic.
Per rectum: normal anal tone, no fissures, stenosis, or hemorrhoids.

Laboratory Findings
Routine hematological and biochemical tests were normal.

Radiological Studies
- Pelviabdominal Ultrasound: normal.
- CT Abdomen and Pelvis: No obstructive lesion, mass, or structural abnormality detected.

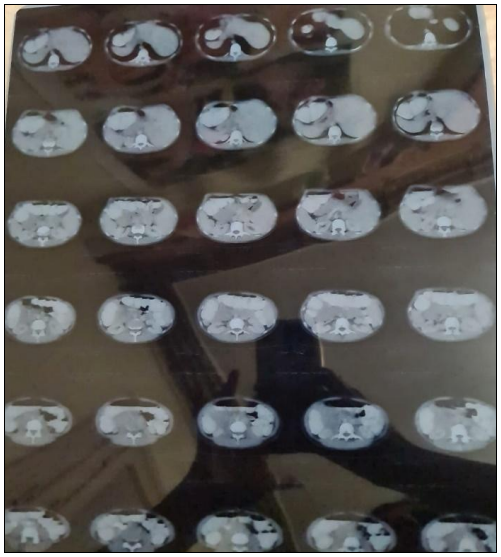

Fig 1: Axial and Coronal CT Abdomen and Pelvis showing no obstructive mass or structural lesion.

Barium enema: short funnel-shaped distal segment with proximal dilatation, indicating a transition zone.

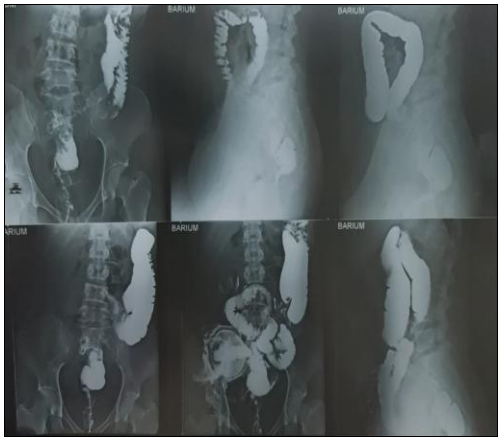

Fig 2: Barium enema showing the transition zone.

MRI lumbosacral spine: No abnormalities detected, excluding neurological causes.

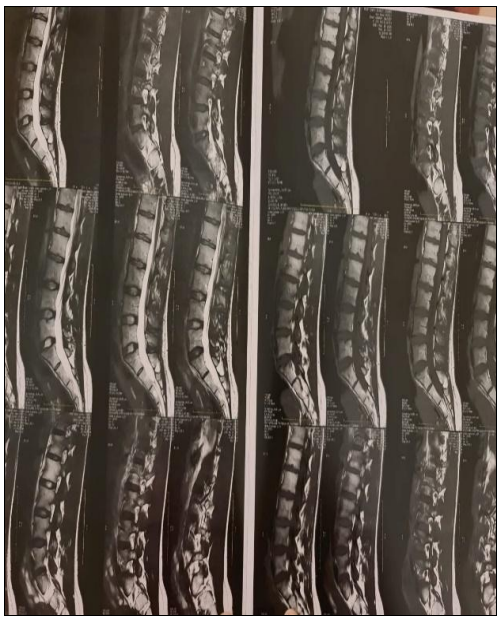

Fig 3: Sagittal and axial MRI of the lumbosacral spine confirming no structural neurological abnormalities contributing to the constipation.

Anorectal Manometry
Findings included increased resting pressure (95 mmHg), elevated rectal sensory thresholds, increased compliance, and paradoxical anal contraction—features consistent with functional outlet obstruction secondary to hypoganglionosis [4].

Preoperative Biopsy
Full-thickness rectal biopsy showed markedly reduced ganglion cells and hypertrophied nerve fibers.

Surgical Management
Exploration revealed a transitional zone at the distal descending colon. The sigmoid and rectum were collapsed distal to a dilated proximal colon. Resection of the sigmoid and rectum was performed with a stapled colo-anal anastomosis and protective loop ileostomy. Recovery was uneventful. The ileostomy was closed after two months, with restoration of normal bowel habits.

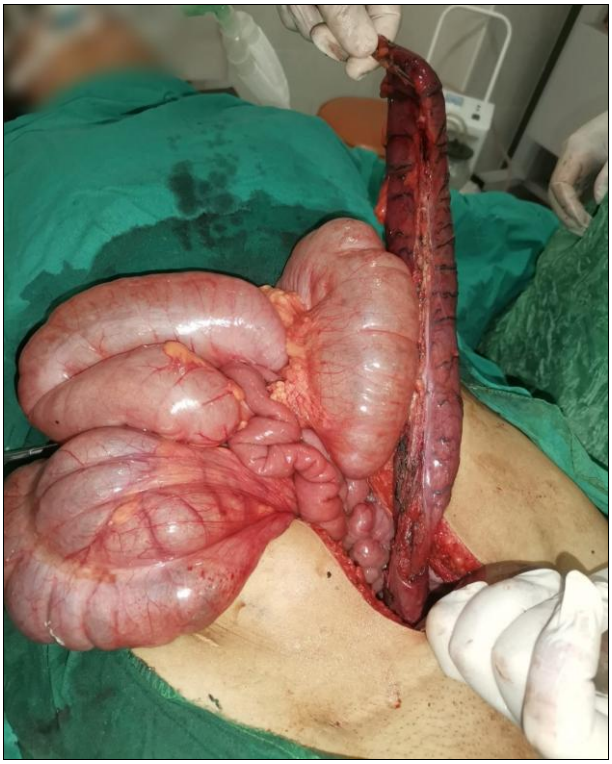

Fig 4: Intraoperative photo showing the dilated proximal colon and the about-to-be resected rectosigmoid colon.

Histopathology
Gross: Dilated proximal segment and narrow distal segment (18 cm).

Microscopy
- Proximal: normal ganglion cells, mild nerve hypertrophy, active bilharzial colitis.
- Distal: markedly reduced ganglion cells (hypoganglionosis), hypertrophied nerve fibers.

Final diagnosis: segmental hypoganglionosis with background bilharzial colitis.

Discussion
Rarity and Diagnostic Delay
Adult-onset hypoganglionosis is rare but clinically significant. Most reports describe delayed diagnosis after years of symptoms [5]. Chronic constipation unresponsive to conventional therapy should prompt consideration of enteric neuropathies.

Pathogenesis: Congenital vs. Acquired
Two mechanisms are proposed: congenital underdevelopment of enteric ganglia with late manifestation, or acquired ganglion loss secondary to inflammatory, ischemic, or immune-mediated injury [6].
In this patient, symptom onset soon after appendectomy suggests an acquired process, possibly triggered by localized inflammation or ischemia [7]. The coexistence of bilharzial colitis supports a secondary insult contributing to ganglionic depletion [8].

Post-Appendectomy Pathophysiologic Links
Possible mechanisms include:
1. **Post-inflammatory ischemic injury:** Surgical procedure “appendectomy” in the right lower quadrant

may have resulted in localized peritonitis or ischemia. These postoperative inflammatory processes could have injured the enteric nervous system (ENS), mainly the myenteric plexus [7].

2. **Immune-mediated ganglionitis** initiated by postoperative inflammation [9].
3. **Bilharzial colitis** inducing chronic transmural inflammation and neuronal damage [8].
4. **Surgical plexus disruption**, altering local motility patterns.
5. **Unmasking of mild congenital hypoganglionosis** following surgical stress [6].

This multifactorial pathogenesis likely explains the patient's progressive course.

Clinical and Radiologic Features

Typical findings include chronic refractory constipation, abdominal distension, and evidence of a transition zone on imaging. Contrast enema and manometry are useful but not diagnostic alone [10].

In our patient contrast enema (funnel-shaped distal segment) and manometry (paradoxical anal contraction) supported a functional obstructive process with a distal transition zone.

Diagnosis and Role of Biopsy

Histopathologic confirmation via full-thickness biopsy is essential [11]. Immunohistochemical stains such as calretinin and AChE aid evaluation of ganglion density.

Surgical Management and Outcomes

Definitive management is resection of the hypoganglionic segment with anastomosis to normal bowel [12]. Outcomes are favorable when resection margins encompass fully ganglionic tissue and a protective stoma is used [13].

Our patient achieved full resolution following sigmoid and rectal resection with colo-anal anastomosis.

All data taken together, the most possible explanation in this patient is a multifactorial pathogenesis, with a postoperative inflammatory or an immune-mediated neuronal injury potentiated by bilharzial enteric inflammation and possibly latent congenital hypoganglionosis. Recognition of acquired or secondary hypoganglionosis is critical, as early surgical intervention tailored to the affected segment can achieve excellent outcomes, as seen in this case.

Differential Diagnosis

Includes adult Hirschsprung disease, colonic inertia, and pelvic floor dyssynergia. Multidisciplinary assessment (surgery, gastroenterology, pathology) ensures accurate diagnosis [14].

Conclusion

Adult-onset rectal hypoganglionosis is an uncommon but surgically curable cause of chronic constipation. Diagnosis requires high suspicion, radiologic evaluation, and full-thickness biopsy. Early surgical resection offers excellent long-term outcomes when the affected segment is fully excised.

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Conflict of Interest

The author declares no conflict of interest.

Ethical Approval: Ethical approval was not required for this case report, as per the institutional policy of Al-Azhar University Hospital, New Damietta, Egypt.

Informed Consent: Written informed consent was obtained from the patient for publication of this case report and the accompanying images.

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