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Case Report
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Neonatal Hypoganglionosis Presenting as Acute Appendicitis in a 3-year-old Girl: A Case Report

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Abstract

Neonatal hypoganglionosis is a rare congenital dysganglionosis typically presenting in the neonatal period with intestinal obstruction. Initial symptoms may include intermittent massive abdominal distension and non-bilious vomiting. Presentation beyond infancy is common, often subtle and easily missed, while acute appendicitis following inadequate conservative therapy is exceptionally rare. We describe a 3-year-old girl who presented with atypical features of acute appendicitis, posing a diagnostic challenge. She underwent laparoscopic appendicectomy, and previously neonatal rectal suction biopsy histopathology revealed features consistent with hypoganglionosis involving the distal rectum. This case highlights the diagnostic complexity of atypical enteric neuropathies and underscores the importance of histological evaluation in neonatal distal partial functional bowel obstruction.

Keywords: Hypoganglionosis; Enteric Neuropathy; Acute Appendicitis; Paediatric Motility Disorder; Fecalith; Histopathology; Appendicectomy; Congenital Dysganglionosis; Hirschsprung Disease Differential; Bowel Regimen; Myenteric Plexus; Delayed Presentation; Neonatal Bowel Dysfunction; Paediatric Gastrointestinal Surgery

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Introduction

Hypoganglionosis is a rare congenital abnormality of the enteric nervous system, characterised by reduced ganglion cell density in the myenteric plexus. It typically presents in the neonatal period with intestinal obstruction, abdominal distension, or failure to pass meconium or stools later in infancy [1].

Late presentations are often subtle, easily overlooked, and may mimic more common conditions such as constipation, pseudo-obstruction, or Hirschsprung disease. Acute appendicitis is a common surgical emergency in children; however, its association with underlying enteric neuropathies is rarely reported [2]. This case illustrates an unusual presentation of congenital hypoganglionosis manifesting as acute appendicitis in a previously diagnosed and conservatively inadequately treated hypoganglionosis 3-year-old girl.

Case Report

A 2200 g female neonate was born to a primigravida at 37 weeks via emergency caesarean section due to intrauterine growth restriction and fetal distress. Baby was in good condition with normal APGAR scores. Urine and meconium were passed soon after birth, and breastfeeding was well established. Both mother and baby were discharged the following day.

In the first two weeks, the baby experienced occasional vomiting in the supine position, which improved with postural adjustments. However, by the third week, she developed sudden and massive abdominal distension with increased frequency and volume of postprandial vomiting. She struggled to pass stools and flatus. The general practitioner, alarmed by the rapid onset of symptoms, referred her urgently for suspected neonatal intestinal obstruction.

On examination, the infant weighed 2340 g and appeared well and playful. The abdomen was globally distended with a tympanic note on percussion. Bowel sounds were mildly exaggerated. No palpable mass or signs of peritonism were present. Hernial sites were normal. Perineal examination

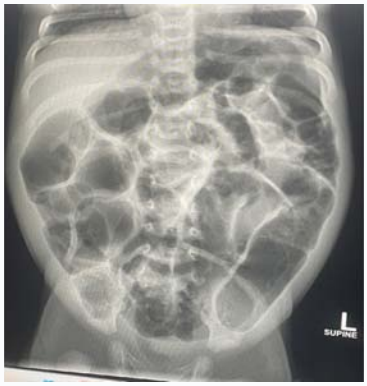

Figure 1: Neonatal abdominal radiograph.
Supine abdominal radiograph obtained at 3 weeks of age demonstrating generalised gaseous distension of bowel loops with gas visible up to the upper rectum, consistent with functional partial distal bowel obstruction. No air–fluid levels or mechanical transition point are seen.

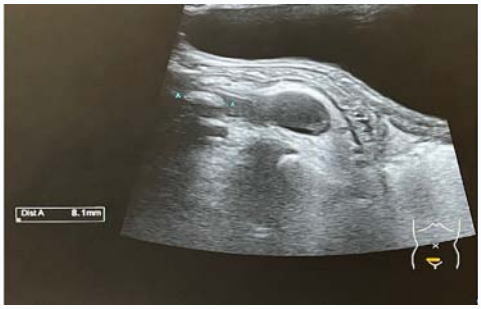

Figure 2: Ultrasound of the right iliac fossa at age 3 years.
Longitudinal ultrasound image showing a non-compressible tubular structure distal dilated part measuring 10 mm in diameter, containing an echogenic fecalith of 8.1 mm casting posterior acoustic shadowing. The distal appendix appears dilated and inflamed, with periappendiceal fluid, consistent with acute appendicitis.

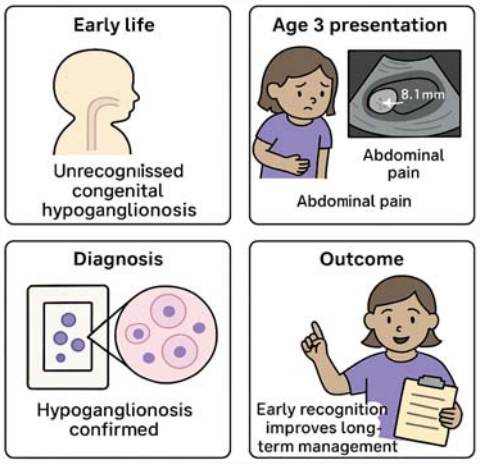

Figure 3: Infographic-style graphical abstract.

Pathophysiological Link

Condition	Mechanism	Impact
Hypoganglionosis	Reduced ganglion cells in the myenteric plexus → impaired colonic motility	Chronic constipation, stasis, segmental dysmotility
Constipation	Prolonged stool retention → increased intraluminal pressure and bacterial overgrowth	Faecalith formation, luminal obstruction
Appendicitis	Obstruction of appendiceal lumen (often by faecalith) → inflammation and infection	Acute abdominal pain, surgical emergency

Figure 4: Conceptual map of the pathophysiological relationship.
Flow-diagram illustrating the sequence from hypoganglionosis → impaired colonic motility → chronic constipation → fecal stasis → fecalith formation → appendicitis.

revealed three distinct openings (urethra, vagina, and anal canal). The anus was normally sited and of appropriate size on calibration. Some flatus was relieved after anal calibration, but the response was not typical of Hirschsprung disease.

Urinalysis, hematology, biochemistry, and C-reactive protein were all within normal limits. Abdominal radiograph showed generalised bowel distension with gas visible up to the upper rectum suggestive of functional partial distal bowel obstruction (Figure 1).

The baby responded well to rectal saline washouts (10 mL/kg, three times daily) and was discharged. However, she required decompression almost twice weekly, and her parents struggled to manage the regimen at home.

Later Presentation at Age 3

At age 3, the child presented with a 24-hour history of right lower quadrant abdominal pain, constipation, and vomiting. Her parents reported intermittent constipation and diarrhea since infancy, managed conservatively. There were no prior hospital admissions or growth concerns. No family history of gastrointestinal or neurological disorders was noted.

On examination, she was afebrile with stable vital signs. Abdominal examination revealed mild, deep tenderness in the lower abdomen, more pronounced on the right. Bowel sounds were present but reduced. No abdominal distension was observed.

Investigations

Urinalysis and blood tests, including inflammatory markers (CRP and neutrophils), were normal.

Ultrasound of the abdomen and pelvis revealed a non-compressible tubular structure in the right iliac fossa measuring 10 mm, containing an 8.1 mm fecalith. There was a small amount of periappendiceal free fluid, with no mass—findings consistent with acute appendicitis.

Differential Diagnosis

Acute appendicitis, mesenteric adenitis, constipation-related abdominal pain, intussusception, early small bowel obstruction, variant Hirschsprung disease, other enteric neuropathies were considered as the possible differential diagnosis. Given the clinical and radiological findings, a diagnosis of acute appendicitis was made.

Treatment

The patient underwent urgent laparoscopic appendicectomy. Intraoperative findings included a large fecalith occupying the proximal two-thirds and distal one-third of the appendix, with inflamed and oedematous distal appendix. No perforation or mechanical obstruction was noted. The postoperative course was uncomplicated, and the patient was discharged on day 2.

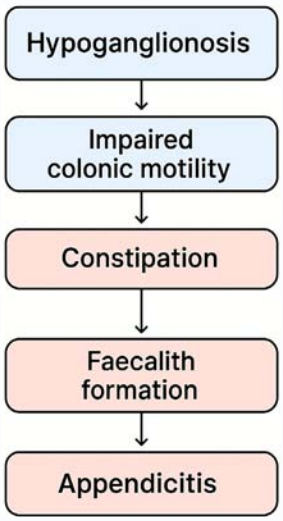

Figure 5: This flowchart illustrates the full pathophysiological sequence from hypoganglionosis to appendicitis, integrating: Histopathology: Reduced ganglion cells, hypertrophic nerve fibers, Neuronal density gradient: ↓ ganglion cells in myenteric plexus, Transition zone: Subtle segmental dysmotility, Constipation and fecal stasis: Leading to fecalith formation, Appendicitis: Final inflammatory outcome It lays out the relationship between hypoganglionosis → impaired motility → constipation → fecalith formation → appendicitis in a clean, abstract medical-illustration style.

Outcome and Follow-Up

Given the conservatively treated hypoganglionosis , the child was referred to paediatric gastroenterology. Further evaluation included:

At 6-month follow-up, she remained well with improved bowel habits on a structured bowel regimen. No further surgical intervention was required.

Discussion

The report highlights several clinically important points. Hypoganglionosis may present in the neonatal period and mimic common paediatric surgical emergencies. Acute appendicitis can be the complication of an underlying enteric neuropathy. Histopathological examination of rectal suction biopsy specimens in neonates and infants can reveal unexpected congenital abnormalities [3].

This case contributes to the limited literature on atypical presentations of enteric dysganglionosis and underscores the importance of multidisciplinary evaluation in children with subtle motility symptoms.

Hypoganglionosis is a rare form of intestinal dysganglionosis characterised by reduced ganglion cell density and abnormal innervation. It is typically classified into congenital and acquired forms, with congenital hypoganglionosis presenting early in life [4].

This case is unusual for several reasons:

1. **Early presentation**: Most cases present late; presentation at 3 weeks is rare.
2. **Appendicitis as complication manifestation**: Appendicitis is a typical presenting feature of inadequately treated hypoganglionosis. Reduced peristalsis and impaired motility may predispose to luminal stasis and inflammation.
3. **Histological diagnosis**: The diagnosis was made early following rectal suction biopsy, emphasising the value of routine histopathology in neonatal partial functional low large bowel obstruction in the absence of any mechanical one.

The overlap between hypoganglionosis and Hirschsprung disease can complicate diagnosis. Unlike Hirschsprung disease, hypoganglionosis shows reduced but not absent ganglion cells, and the transition zone may be subtle.

Management depends on severity. Many children require bowel management programs, while severe cases may need endosurgical procedures, Botox injections or surgical resection. Our patient responded well to conservative management [5].

The relationship between hypoganglionosis, constipation, and appendicitis is subtle but clinically significant. Here's a synthesis of current understanding based on recent case reports and literature: Figure 4 and 5.

How They Interact

Hypoganglionosis leads to chronic constipation due to impaired peristalsis and segmental colonic dysmotility. Constipation increases the risk of fecalith formation, especially in the appendix, where stasis is common. Fecaliths obstruct the appendiceal lumen, triggering inflammation, bacterial invasion, and ultimately acute appendicitis [6].

Case-Based Insights

- Adult-onset hypoganglionosis following appendectomy -A 27-year-old woman developed refractory constipation after appendectomy, later diagnosed with rectal hypoganglionosis. Relevance: Demonstrates the link between hypoganglionosis, chronic constipation, and appendiceal pathology. Supports the concept that dysmotility can contribute to appendicitis or appendicitis-like presentations.
- A 12-year-old boy with achalasia and severe constipation was found to have acquired myenteric hypoganglionosis after surgical complications.
- In another case, abdominal compartment syndrome developed due to massive fecal loading in hypoganglionosis, requiring emergency colectomy.
- Hypoganglionosis mimicking appendicitis in a toddler. Relevance: A toddler with known hypoganglionosis initially presented with symptoms mimicking appendicitis. Supports your case's clinical plausibility.
- Hypoganglionosis presenting as appendicular mass. Relevance: Highlights how hypoganglionosis can mimic appendiceal disease, reinforcing the diagnostic overlap.

How these references support our case

They collectively show that hypoganglionosis can present with abdominal pain, peritonism, or appendicitis-like symptoms, even though true appendicitis is rare. They support the pathophysiological link between chronic constipation, fecal stasis, fecalith formation, and appendiceal inflammation. They demonstrate that histopathology is essential for diagnosing underlying enteric neuropathies [7].

Clinical Implications

Children with chronic constipation and appendicitis may have underlying enteric neuropathies like hypoganglionosis. Histopathological examination of rectal suction biopsy specimens

can reveal unexpected diagnoses. Early recognition allows for tailored bowel management and prevents recurrent surgical emergencies. Hypoganglionosis leads to impaired motility and chronic constipation, which increases the risk of fecal stasis and fecalith formation. Fecaliths are a well-recognised cause of appendiceal obstruction and inflammation. Paediatric cases of hypoganglionosis mimicking appendicitis have been reported, including toddlers presenting with peritonism and abdominal pain [8].

Learning Points

- Hypoganglionosis is a rare congenital enteric neuropathy that may present in infancy.
- Acute appendicitis can be an unusual manifestation of underlying motility disorders.
- Histopathological examination of rectal suction biopsy specimens in children can reveal unusual diagnosis other than Hirschsprung's disease.
- Chronic constipation increases the risk of fecalith-related appendicitis.
- Early recognition of enteric neuropathies allows timely referral and long-term bowel management.

Conclusion

This case demonstrates the complex and evolving nature of congenital enteric neuropathies and highlights how neonatal hypoganglionosis may remain clinically silent or only partially responsive to conservative management, later manifesting as an atypical surgical emergency. The progression from subtle neonatal functional obstruction to fecalith-associated acute appendicitis at age three underscores the importance of maintaining diagnostic vigilance in children with long-standing bowel dysfunction. Histopathology played a pivotal role in establishing the underlying diagnosis when symptoms are atypical or disproportionate to imaging findings. Early recognition of enteric neuropathies enables timely referral, structured bowel management, and prevention of recurrent complications. This case emphasises that even when initial neonatal symptoms appear to resolve, underlying dysganglionosis may persist and present unexpectedly years later.

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