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Case Report

Coexistence of achalasia cardia and Hirschsprung's disease in a child: a rare case report

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ABSTRACT

Achalasia cardia and Hirschsprung's disease are distinct pediatric gastrointestinal motility disorders that rarely manifest in a single patient. While both involve a failure of muscular relaxation within the enteric nervous system, they typically present with disparate clinical features. We report an exceedingly rare case of a 3-year-old girl who initially presented with classic signs of achalasia cardia, including progressive dysphagia and regurgitation. Following a successful Heller's cardiomyotomy, the patient's esophageal symptoms improved; however, her recovery was hampered by emerging constipation and significant abdominal distension. Initially attributed to protein-energy malnutrition, these lower gastrointestinal symptoms prompted further investigation, which ultimately revealed a concurrent diagnosis of Hirschsprung's disease. The patient subsequently underwent a posterior anal myectomy, which led to the complete resolution of her symptoms and a full recovery. This case underscores the diagnostic complexity of dual motility pathologies. It emphasizes that persistent or evolving symptoms following a primary surgical intervention should prompt a high index of suspicion for additional underlying conditions, requiring a comprehensive and multidisciplinary diagnostic approach to ensure favorable pediatric outcomes.

Keywords: achalasia cardia, Hirschsprung's disease, pediatric GI disorders, Heller's cardiomyotomy, anal myectomy

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Introduction

Achalasia cardia is an uncommon esophageal motility disorder characterized by degeneration of neurons within the myenteric plexus, leading to impaired relaxation of the lower esophageal sphincter and uncoordinated esophageal peristalsis [1]. Though more common in adults, it can occur in children and is often misdiagnosed due to overlapping symptoms with other gastrointestinal disorders [2]. Hirschsprung's disease is a congenital enteric neuropathy in which ganglion cells are absent in segments of the distal intestine, resulting in impaired bowel motility and functional intestinal obstruction [3].

Although both conditions involve neuronal dysfunction of the digestive tract, they differ in pathophysiology and clinical presentation. The simultaneous occurrence of achalasia and Hirschsprung's disease is exceedingly rare and poses a diagnostic challenge [4,5]. We report such a case to highlight the importance of thorough evaluation in pediatric patients' persistent or evolving gastrointestinal symptoms. This case adds to the limited global literature and emphasizes the importance of considering coexisting enteric neuropathies in persistent pediatric GI dysfunction

Case Report

A 3-year-old female presented with a six-month history of progressive dysphagia to both solids and liquids, associated with regurgitation of undigested food and failure to thrive. On presentation, she appeared chronically ill with evidence of poor nutritional status, and anthropometric assessment was consistent with failure to thrive for age. Laboratory investigations were within acceptable limits for surgery.

A contrast esophagogram demonstrated a markedly dilated esophagus with smooth tapering ("birdbeak" appearance) at the gastroesophageal junction and delayed esophageal emptying, findings highly suggestive of achalasia cardia. Based on the characteristic clinical presentation and radiologic features, a working diagnosis of achalasia was made. Esophageal manometry, which remains the diagnostic gold standard for confirming achalasia, was not available in our center at the time of evaluation and therefore could not be performed.

This limitation is acknowledged; however, in resource-

limited settings, the combination of typical symptoms and classical radiologic findings on contrast esophagography is often relied upon to establish the diagnosis. The patient was optimized nutritionally and medically before undergoing a modified open Heller's cardiomyotomy, with initial postoperative relief of symptoms. (Figure 1).



Figure 1. Open Modified Heller's cardiomyotomy via thoracotomy.

However, within a few weeks, she developed progressive constipation and abdominal distension. These symptoms were initially misdiagnosed as protein-energy malnutrition and managed conservatively.

As the symptoms persisted, further evaluation with a contrast enema demonstrated a narrowed distal rectal segment with proximal colonic dilatation, suggestive of a transition zone. A provisional diagnosis of Hirschsprung's disease was made.

Six months after the cardiomyotomy, the patient underwent a diverting colostomy with procurement of a full-thickness rectal biopsy taken approximately 2 cm above the dentate line. Histopathological examination using hematoxylin and eosin staining demonstrated absence of ganglion cells within the submucosal and myenteric plexuses, with associated hypertrophy of submucosal nerve fibers, confirming aganglionosis consistent with Hirschsprung's disease.

A posterior anal myectomy was performed several months later. One year after the initial presentation, the patient subsequently underwent colostomy takedown following satisfactory postoperative recovery and improvement in bowel function.

Following the staged surgical procedures, the patient achieved complete resolution of her gastrointesti-

nal symptoms. Six months after definitive surgery, she demonstrated regular bowel habits, satisfactory weight gain, and evidence of catch-up growth. Continued follow-up confirmed sustained clinical improvement and a satisfactory quality of life. Informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Discussion

Achalasia and Hirschsprung's disease are rare but significant pediatric digestive tract disorders with overlapping clinical features yet distinct pathophysiological mechanisms. Achalasia cardia is characterized by failure of the lower esophageal sphincter (LES) to relax and, absence of esophageal peristalsis, primarily due to loss of inhibitory ganglion cells in the myenteric (Auerbach's) plexus [6]. Hirschsprung's disease similarly results from a lack of ganglion cells, but in the distal colon and rectum, leading to chronic functional intestinal obstruction [3].

While achalasia is commonly recognized in adults, pediatric achalasia is far less common and often underdiagnosed or misdiagnosed [7]. Pediatric achalasia accounts for less than 5% of all achalasia cases [8]. The annual incidence in children is estimated at 0.11 per 100,000 children, with a peak presentation age between 8 and 12 years, though cases in younger children and infants have been reported [9].

Achalasia may occur as part of the rare AAA syndrome, also referred to as Allgrove syndrome, which is characterized by the triad of alacrima, achalasia, and adrenal insufficiency (Addison disease). This condition results from mutations in the AAAS gene located on chromosome 12q13, which encodes the ALADIN protein involved in nuclear pore complex function. These manifestations are incorporated into the Eckardt scoring system, a clinical tool widely used to assess symptom severity and treatment outcomes in patients with achalasia [10].

In young children, the clinical presentation of achalasia is often nonspecific and may mimic other conditions such as gastroesophageal reflux disease (GERD), esophageal stricture, or feeding aversion disorders. Affected patients commonly present with progressive dysphagia, regurgitation of undigested food, recurrent vomiting, chronic cough, weight loss, and failure to

thrive. Retrosternal chest discomfort may also occur in older children and adults. Because these symptoms develop gradually and overlap with more common pediatric gastrointestinal conditions, the diagnosis is frequently delayed.

The diagnosis is typically established using a combination of barium swallow (which reveals the classic "bird-beak" sign), esophageal manometry (demonstrating incomplete LES relaxation and aperistalsis), and upper gastrointestinal endoscopy to rule out structural causes. In many low-resource settings, as in our case, diagnosis is made using barium swallow studies due to limited access to manometry and endoscopic equipment [11].

Treatment options for achalasia in children include Heller's cardiomyotomy (open or laparoscopic), pneumatic balloon dilation, and, more recently, peroral endoscopic myotomy (POEM), with Heller's myotomy remaining the standard surgical approach in resource-limited settings. The long-term outcomes in pediatric patients undergoing

Heller's myotomy is generally favorable, with improvement in dysphagia and weight gain reported in up to 90% of cases [12].

Hirschsprung's disease, in contrast, is a congenital condition typically diagnosed in the neonatal period or early infancy [13]. However, in ultra-short segment or atypical cases, diagnosis may be delayed until later in childhood [14]. Common symptoms include delayed passage of meconium, chronic constipation, abdominal distension, and failure to thrive. Diagnosis is confirmed by rectal biopsy demonstrating absence of ganglion cells. A contrast enema revealing a transition zone between aganglionic and ganglionic bowel segments supports the diagnosis [7].

The simultaneous presence of achalasia and Hirschsprung's disease has been reported only rarely in the medical literature [4]. Only a handful of cases have been reported in the literature [5]. The shared involvement of the enteric nervous system in both conditions has prompted speculation about a common embryological or genetic origin. Some researchers have postulated that both disorders may fall within the spectrum of neurocristopathies diseases arising from aberrant development of neural crest cells. Mutations in genes such as RET, EDNRB, and SOX10, which are implicated in the develop-

ment of the enteric nervous system, have been associated with Hirschsprung's disease and may also play a role in esophageal motility disorders [14,15]. However, definitive genetic or mechanistic links remain unclear due to the rarity of co-presentation and limited data.

Our case exemplifies the diagnostic difficulty in identifying dual pathologies, particularly when the second condition presents shortly after surgical correction of the first. In this case, postoperative constipation and abdominal distension were initially attributed to malnutrition and reduced motility following Heller's myotomy. The delay in diagnosing Hirschsprung's disease could have been avoided with earlier consideration of dual pathology, especially in the presence of hallmark signs such as persistent constipation and a distended abdomen. This diagnostic oversight has been reported in other pediatric cases, underscoring the need for vigilance [16,17].

Surgical management of Hirschsprung's disease typically involves a pull-through procedure (e.g., Soave or Duhamel), although limited distal disease may be treated with posterior anal myectomy, as was done in this patient. This approach also provides tissue for histological confirmation of aganglionosis.

The favorable outcome following staged surgical management underscores the importance of maintaining a high index of suspicion for dual pathology in children with persistent or unexplained symptoms despite initial treatment. Timely diagnostic reassessment can significantly improve outcomes. Awareness of such rare coexisting conditions is essential to avoid diagnostic delays and ensure appropriate surgical intervention.

In conclusion, this report emphasizes the importance of considering coexisting digestive tract pathologies in pediatric patients with persistent or atypical symptoms. Early diagnosis and multidisciplinary management are key to optimizing outcomes. The rare combination of achalasia and Hirschsprung's disease should be considered in complex pediatric gastrointestinal presentations.

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Authors' contribution

All authors contributed to the conception, data collection, writing, and final approval of the manuscript.

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