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Case Report

Association of achalasia and Down syndrome in an adult patient: a case report

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ABSTRACT

Achalasia is a rare primary esophageal motility disorder caused by degeneration of inhibitory neurons in the myenteric plexus, resulting in impaired lower esophageal sphincter (LES) relaxation and absent peristalsis. Adult-onset achalasia in individuals with Down syndrome is exceedingly uncommon and may be underrecognized due to subtle or atypical symptom expression. This case, reported adult with Down syndrome and achalasia, highlights the importance of early recognition and multidisciplinary care. We report a 63-year-old man with genetically confirmed Down syndrome presenting with progressive dysphagia to solids, postprandial regurgitation, intermittent retrosternal discomfort, and a 10 kg weight loss over six months. Oral intake was markedly restricted, and communication limitations hindered symptom reporting. Upper endoscopy revealed retained semi-solid food and resistance at the LES without mucosal lesions. High-resolution manometry (HRM) confirmed type II achalasia with 100% failed peristalsis and panesophageal pressurization. The patient underwent laparoscopic Heller myotomy with anterior (180°) Dor fundoplication. Intraoperative findings included tissue laxity, gastroesophageal junction fibrosis, and a 1.5 cm hiatal hernia, which was repaired. Recovery was uneventful. At one-year follow-up, the patient achieved complete resolution of dysphagia, normalized oral intake, sustained weight gain (BMI 17.8 to 23.1 kg/m²), and absence of reflux. Adult-onset achalasia in Down syndrome is rare but clinically significant. Diagnosis and timely laparoscopic intervention provide durable symptom relief and nutritional improvement. This case emphasizes the importance of clinical vigilance and multidisciplinary care for optimal outcomes in this population.

Keywords: Achalasia, Down syndrome, Esophageal motility disorder, Heller myotomy, Dor fundoplication, Nutritional recovery, Type II achalasia

INTRODUCTION

Achalasia is a rare primary esophageal motility disorder characterized by impaired relaxation of the lower esophageal sphincter (LES) and absent or ineffective esophageal peristalsis, leading to functional obstruction, progressive dysphagia, regurgitation, and weight loss.[1-3] Its estimated incidence is approximately 1 per 100,000 individuals per year, with peak presentation between 25 and 60 years of age, and no significant sex predilection.[2,4]

Familial clustering is uncommon, though sporadic genetic variations may contribute to disease susceptibility.[4,5] Delayed diagnosis is associated with chronic esophageal dilation, malnutrition, aspiration, and increased morbidity, underscoring the importance of early recognition and intervention.

Pathophysiologically, achalasia results from progressive degeneration of inhibitory neurons within the myenteric plexus (Auerbach), causing an imbalance between excitatory and inhibitory signaling, impaired LES relaxation, and disordered esophageal peristalsis.[1,5] Etiological mechanisms are multifactorial and include autoimmune processes, viral-mediated injury, and chronic inflammation.[4,5] Emerging studies suggest immune-mediated injury contributes to esophageal neuronal degeneration in genetically predisposed individuals.[4,5] Such molecular insights may explain the increased vulnerability of certain populations, including patients with Down syndrome, to esophageal motility disorders.

Individuals with Down syndrome (trisomy 21) are predisposed to a variety of gastrointestinal disorders, including gastroesophageal reflux disease (GERD), chronic constipation, and Hirschsprung disease, largely due to enteric nervous system anomalies, hypotonia, and autonomic dysregulation.[6,9] Esophageal dysmotility is well documented in pediatric and adolescent populations, but adult-onset achalasia is exceedingly rare, with fewer than 20 cases reported in the literature.[9,11] Even asymptomatic adults may present with impaired peristalsis, incomplete LES relaxation, and delayed esophageal emptying, highlighting the need for heightened clinical suspicion, particularly in patients with unexplained dysphagia, regurgitation, or weight loss.[9,12]

High-resolution manometry (HRM) and upper endoscopy are essential for accurate subtype classification and differentiation from pseudoachalasia or secondary motility disorders.[12,13]. According to the Chicago Classification version 4.0, achalasia subtypes include type I, with minimal esophageal pressurization and 100% failed peristalsis; type II, characterized by panesophageal pressurization in ≥20% of swallows with absent peristalsis; and type III, exhibiting spastic contractions with abnormal peristalsis.[2,5] Genetic polymorphisms affecting nitric oxide signaling and neurotransmission may further exacerbate esophageal dysmotility in patients with trisomy 21.[4]

Given the rarity of adult-onset achalasia in Down syndrome, reporting such cases is critical to improve understanding of clinical presentation, diagnostic strategies, and therapeutic outcomes. Here, we present a case of type II achalasia in a 63-year-old man with Down syndrome, highlighting the diagnostic utility of HRM and endoscopy, as well as surgical management through laparoscopic Heller myotomy with Dor fundoplication.[13]

This report emphasizes the importance of early recognition and a multidisciplinary approach in this unique patient population

CASE REPORT

A 63-year-old man with genetically confirmed Down syndrome (47, XY, +21) and psychomotor development consistent with his syndrome presented with progressive dysphagia to solids over six months. Symptoms were accompanied by postprandial regurgitation, intermittent retrosternal discomfort, and a 10 kg weight loss. Oral intake was severely restricted, and communication difficulties limited symptom description. Initial empiric treatment with proton pump inhibitors for presumed GERD did not provide relief.

The patient had no prior abdominal surgeries, endoscopic interventions, smoking or alcohol history, or relevant family gastrointestinal disorders. Physical examination revealed hypotonia, characteristic craniofacial features of Down syndrome, and normal vital signs. Abdominal examination was unremarkable.

Anthropometric assessment showed a weight of 40 kg, height 1.50 m, and Body Mass Index (BMI) of 17.8 kg/m², consistent with malnutrition secondary to chronic dysphagia. Laboratory evaluation revealed mild hypoproteinemia and borderline iron deficiency, without other significant abnormalities.

Upper endoscopy revealed retained semi-solid food in the distal esophagus and resistance at the LES, with no mucosal lesions, strictures, or ulcerations (Figure 1).

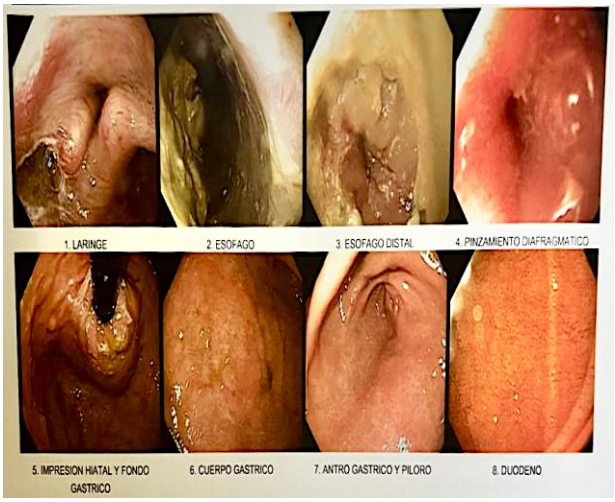

Figure 1: Upper endoscopy showing retained semi-solid food and resistance at the LES, indicative of achalasia.

Biopsies excluded eosinophilic esophagitis and malignancy, suggesting a functional obstruction. HRM demonstrated incomplete LES relaxation (integrated relaxation pressure 32 mmHg), absent peristalsis in 100% of swallows, and panesophageal pressurization in 80% of swallows, consistent with type II achalasia per Chicago Classification v4.0 (Figure 2).

The upper esophageal sphincter was mildly hypertonic, suggesting diffuse neuromotor involvement. HRM was critical in differentiating primary achalasia from pseudoachalasia and secondary motility disorders.

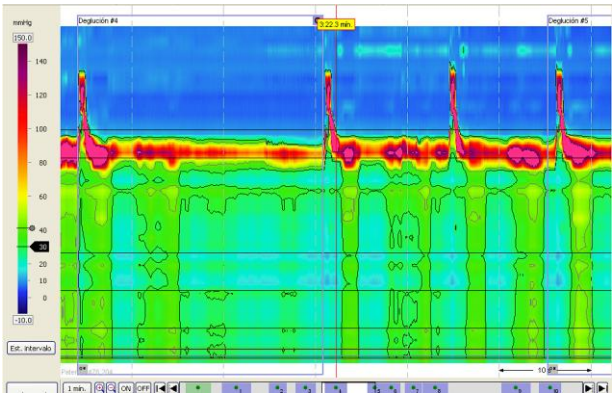

Figure 2: HRM demonstrating 100% failed peristalsis with 80% panesophageal pressurization, consistent with type II achalasia.

The patient underwent laparoscopic Heller myotomy with anterior (180°) Dor fundoplication. Intraoperative findings included increased tissue laxity, fibrosis at the gastroesophageal junction, and a 1.5 cm hiatal hernia, which was repaired. The myotomy extended 5 cm proximally along the esophagus and 2 cm distally onto the gastric cardia using a harmonic scalpel. Intraoperative endoscopy confirmed mucosal integrity and unobstructed passage. Dor fundoplication was applied to reduce postoperative reflux while preserving esophageal emptying.

Post-operative course and follow-up

Recovery was uneventful. Oral intake was advanced gradually from liquids to soft solids. At six months, the patient gained 7 kg, reaching 47 kg (BMI 20.9 kg/m²). At one-year follow-up, weight was 52 kg (BMI 23.1 kg/m²), with complete resolution of dysphagia and reflux. The patient continued multidisciplinary follow-up with gastroenterology, nutrition, and speech-language pathology for ongoing swallowing assessment. Nutritional optimization and monitoring played a key role in functional recovery.[13,15]

This case demonstrates the importance of early recognition, HRM-guided diagnosis, and laparoscopic Heller myotomy with Dor fundoplication in adults with Down syndrome and achalasia. Objective functional assessment, nutritional support, and comprehensive multidisciplinary care were critical to achieving favorable outcomes in this rare patient population.

DISCUSSION

Adult-onset achalasia in Down syndrome is exceedingly rare, with most reports describing pediatric populations and generally favorable postoperative outcomes [8,10,11]. In adults, chronic esophageal stasis may lead to severe dilation, malnutrition, aspiration, and pulmonary complications, emphasizing the importance of timely recognition and intervention.[3,9] This case expands the limited evidence on adult Down syndrome-associated achalasia and highlights clinical and management considerations unique to this population.

The pathophysiology is multifactorial. Histopathological studies reveal enteric nervous system alterations, including neuronal hypoganglionosis, esophageal hypotonia, and neuromuscular dysfunction, which predispose to motility impairment and functional obstruction.[6-9] Immune dysregulation in Down syndrome may increase susceptibility to autoimmune or viral injury targeting the myenteric plexus, promoting progressive neuronal degeneration.[5] Genetic polymorphisms affecting nitric oxide signaling and neurotransmission may further exacerbate esophageal dysmotility.[4] These mechanisms explain the incomplete LES relaxation, absent peristalsis, and atypical clinical presentation, often with markedly reduced oral intake and difficulty expressing dysphagia.

Clinical manifestations in adults with Down syndrome may be subtle due to cognitive or communication limitations. Multiphase swallowing abnormalities, including oropharyngeal involvement, increase aspiration risk. Comprehensive evaluation with HRM and endoscopy, as performed in this case, is essential for accurate subtype classification and for excluding pseudoachalasia or other secondary disorders.[12,13] While a barium esophagram is often recommended, HRM and endoscopic assessment provided sufficient diagnostic clarity to enable timely surgical intervention.

Type II achalasia demonstrates high therapeutic success, exceeding 90% with Heller myotomy or pneumatic dilation, particularly when managed promptly.[4,15] Laparoscopic Heller myotomy with Dor fundoplication is preferred in adults due to its minimally invasive approach, reduced postoperative pain, and preservation of the anti-reflux barrier.[5,14] Intra-operative endoscopic guidance is crucial to minimize mucosal perforation, particularly in cases with fibrosis or hiatal hernia, as demonstrated here.

Emerging minimally invasive therapies, such as peroral endoscopic myotomy (POEM), offer alternative treatment options, though long-term outcomes in Down syndrome populations remain underreported. In this case, laparoscopic Heller myotomy was chosen for its established safety profile and ability to address associated hiatal hernia.

Multidisciplinary care, including gastroenterology, nutrition, and speech-language pathology, is vital to optimize outcomes, prevent malnutrition, and reduce aspiration risk. This patient achieved sustained weight gain, complete symptom resolution, and functional recovery.

Limitations include the single-case design and lack of follow-up HRM or endoscopy. Future studies should aim to define incidence, natural history, and optimal management strategies in adults with Down syndrome, as

well as explore genetic, immunological, and postoperative esophageal remodeling mechanisms.

CONCLUSION

Adult-onset achalasia in Down syndrome is rare but clinically significant, and HRM-guided diagnosis with timely laparoscopic Heller myotomy and partial fundoplication provides safe and effective treatment, yielding durable symptom relief and improved nutritional status. Pathophysiology is multifactorial, involving enteric nervous system abnormalities, immune dysregulation, and potential genetic susceptibility. Early recognition, multidisciplinary management, and individualized care are key to achieving optimal functional outcomes. This case, among the oldest reported adults with Down syndrome and achalasia, underscores the importance of clinical vigilance, comprehensive evaluation, and coordinated care. Further research is needed to clarify incidence, underlying mechanisms, and long-term outcomes in this population.

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