Received: 26 June 2024 | Accepted: 26 November 2024

DOI: 10.1002/jpr3.12161

CASE REPORT

Gastroenterology

Familial polyposis syndrome and achalasia in a young adult

Gabriella A. Lorusso Vivas[1,2] | Ann-Christina Brady[3] | Alejandro Llanos-Chea[1,4]

[1]Department of Pediatrics, Miller School of Medicine, University of Miami, Miami, Florida, USA

[2]Division of Neonatology, Department of Pediatrics, LAC+USC Medical Center, Keck School of Medicine, University of Southern California, Los Angeles, California, USA

[3]Division of Pediatric Surgery, DeWitt-Daughtry Department of Surgery, Miller School of Medicine, University of Miami, Miami, Florida, USA

[4]Division of Pediatric Gastroenterology, Hepatology & Nutrition, University of Texas Southwestern, Dallas, Texas, USA

Correspondence

Alejandro Llanos-Chea, Division of Pediatric Gastroenterology, Hepatology & Nutrition, University of Texas Southwestern, Dallas, TX, USA.
Email: alejandro.llanoschea@utsouthwestern.edu

Funding information
None

Abstract

We report the case of a 19-year-old White male diagnosed with familial adenomatous polyposis (FAP), Gardner's syndrome (GS) phenotype, status post total colectomy, who developed progressive dysphagia and weight loss. He was diagnosed with achalasia based on imaging and esophageal manometry. The patient underwent a Heller myotomy with the resolution of symptoms. To date, no previous literature has reported on concurrent FAP or GS and achalasia. Although FAP and achalasia are both rare conditions with no previously known concurrent occurrence, association, or described syndrome, it is important to be vigilant of this new case report finding. One must also consider the increased risk of malignancy in patients with achalasia in addition to the significant risk of malignancy in patients with FAP/GS.

KEYWORDS

esophageal motility disorder, Gardner's syndrome, polyps

1 | INTRODUCTION

Familial adenomatous polyposis (FAP) and its variants are caused by germline pathogenic variants in the tumor suppressor gene *adenomatous polyposis coli* (*APC*), located on chromosome 5q21-q22. FAP is characterized by hundreds to thousands of colonic adenomatous polyps that often arise in the second and third decades of life. Colon cancer is anticipated if the colon is not removed. Inheritance is autosomal dominant with near complete penetrance of the gastrointestinal (GI) phenotype but with variable penetrance of the extraintestinal manifestations of the disease.[1]

One of the subsets of FAP, Gardner syndrome (GS), presents with additional extraintestinal manifestations such as benign growths, including osteomas and dental abnormalities, cutaneous lesions, congenital hypertrophy of the retinal pigment epithelium, adrenal tumors, and nasal angiofibromas, among others.[2]

Achalasia results from selective degeneration of inhibitory neurons of the esophageal myenteric plexus, which are needed for peristalsis of the smooth muscle of the esophageal body, as well as relaxation of the tonic lower esophageal sphincter.[3] Achalasia has not previously been reported to occur in patients with FAP and/or GS.

We report the case of a young adult male with GS who developed achalasia.

2 | CASE REPORT

We present a 19-year-old White male with GS with a 2-year history of progressive regurgitation of food, heartburn, nausea, and vomiting associated with pharyngeal

dysphagia and unintentional weight loss. His family history was significant for multiple family members affected by FAP. At 9 years of age, he underwent *APC* gene sequencing and deletion duplication analysis, which illustrated a missense mutation in exon 15 (c.239T>A/p.Y799X) consistent with the diagnosis of FAP. No other family members had undergone genetic testing. He underwent multiple upper and lower endoscopies with findings of low-grade dysplasia in his intestinal biopsies.

Initially, his treatment consisted of diet change and pantoprazole for his upper GI symptoms with minimal improvement. Then, one year after the onset of the initial symptoms, due to the presence of tubular adenomas on colonoscopy, he underwent a total colectomy with J-pouch ileoanal anastomosis and protective loop ileostomy. By the time of his surgery, he had no lower GI symptoms related to his FAP. He also underwent routine upper endoscopy as part of his routine FAP surveillance, with findings of mild esophagitis with no gross anatomic abnormality or strictures.

During subsequent months, the patient continued with symptoms of worsening heartburn and dysphagia. Four months after his colectomy, he reported a decreased intake of solids and liquids with an associated weight loss of 7 kg.

An esophagram was performed demonstrating "some contrast transited from the esophagus into the stomach through a very narrow channel in the gastroesophageal junction ("bird's beak" appearance)" (Figure 1). High-resolution esophageal manometry with impedance found panesophageal pressurization, lack of lower esophageal sphincter relaxation with high integrated relaxation pressure (Figure 2), and low baseline impedance with poor variability indicative of the presence of liquid in the esophagus. Those findings were diagnostic for Type II achalasia as per the Chicago v3.0 classification.[4]

The patient underwent a laparoscopic Heller myotomy with partial fundoplication without any complications. Postoperatively the patient received a proton pump inhibitor due to symptoms of reflux. Two months after his procedure, the patient denied any upper GI symptoms, tolerated a regular diet, and regained the lost weight.

3 | DISCUSSION

To our knowledge, this is the first reported case of achalasia in a patient with FAP and/or GS.

FAP and GS are genetic disorders with defined clinical presentations. Their gene mutations and variants have been largely identified.[5] Conversely, the etiology of primary or idiopathic achalasia is unknown. It has been suggested that this disease may be an autoimmune disorder; or there may be a genetic predisposition to the inflammatory degeneration of ganglion cells. It has also been suggested there may be an allergy-driven form of achalasia.[6]

Achalasia can also be secondary, also known as pseudoachalasia, with multiple causes reported (Table 1).[6] Within these causes, FAP, GS, or any other polyposis syndrome have not been reported. Given the nature of this case report, we cannot define an association between both conditions. We know that FAP/GS has a genetic base,[1,2,5] and achalasia has been proposed to have a potential genetic predisposition.[6] Nevertheless, to date, no FAP/GS genes have been described in patients with achalasia.

There are no curative options for achalasia. Treatments for achalasia are aimed to improve the quality of life, preserve esophageal function, and prevent esophageal stasis. These include medications, botulinum toxin injection to the lower esophageal sphincter, pneumatic balloon dilation, and surgical myotomy with or without an anti-reflux procedure. Pneumatic dilatation and myotomy are the preferred and most effective treatment options. Myotomy can be done by laparoscopy or endoscopically (peroral endoscopic myotomy).[7]

The increased risk for malignancy in FAP and GS is well known. Achalasia also increases the risk for esophageal malignancy, suspected to be caused by retention esophagitis or chronic reflux following intervention.[8] Therefore, it is essential to communicate with patients and minimize additional malignancy risks.

The quality of life for patients with conditions such as FAP, GS, or achalasia is dependent on the appropriate and timely management of symptoms, the success of

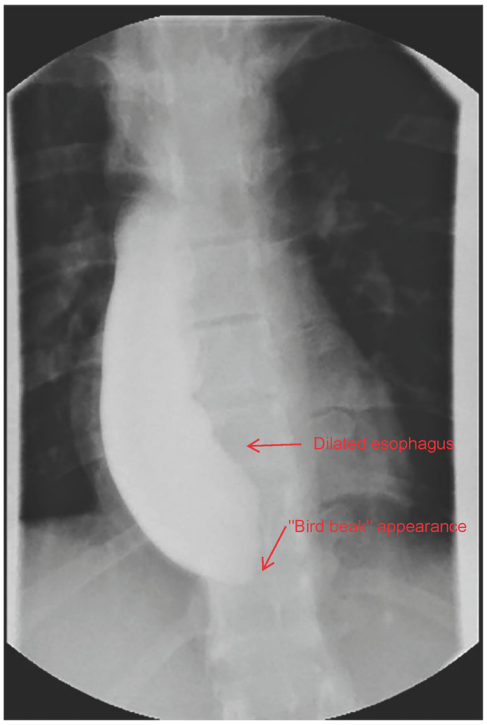

FIGURE 1 Barium esophagram with "bird's beak appearance" of the distal esophagus.

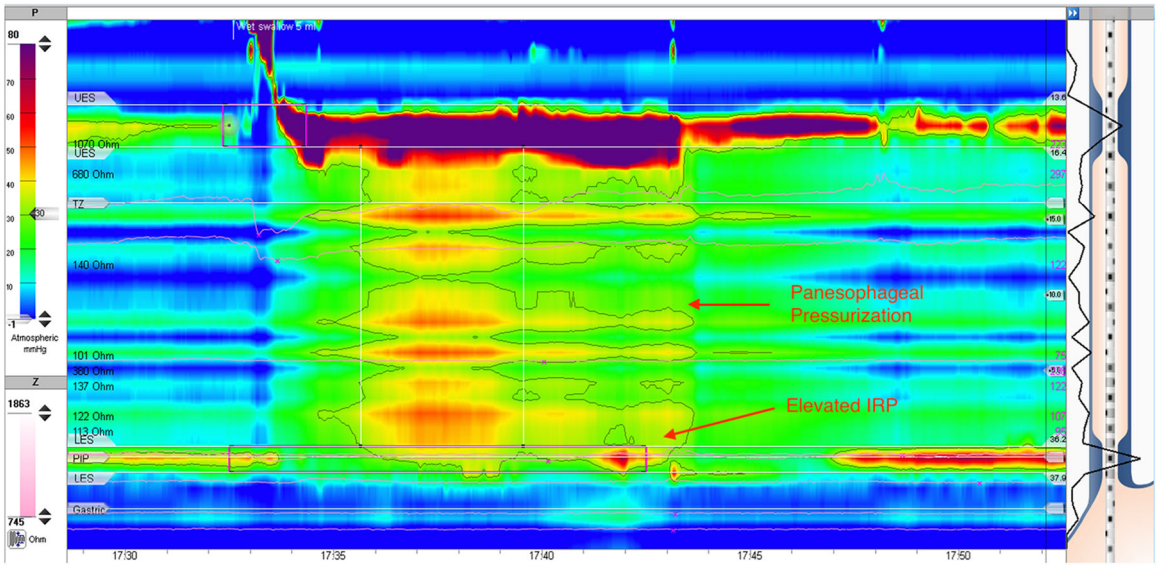

FIGURE 2 Esophageal manometry with absent lower esophageal sphincter relaxation and panesophageal pressurization.

TABLE 1 Secondary causes of achalasia.[6]

Secondary causes of achalasia	
Benign	**Malignant**
• Chagas disease	• Metastatic disease
• Eosinophilic esophagitis	• Paraneoplastic disease
• Postsurgical (fundoplication, gastric banding, truncal vagotomy, gastrectomy, thoracic endovascular aneurysm repair)	• Primary malignancy of the stomach, esophagus, or gastro-esophageal junction
• Esophageal mesenchymal tumors	
• Neurological disorders	
• Pancreatic pseudocyst	
• Peritoneal sclerosis	
• Descending aorta aneurysm	
• Histiocytosis X	
• Mediastinal fibrosis	
• Amyloidosis	
• Sarcoidosis	

therapeutic interventions, and the reduction of cancer risk. Individualized management should be tailored for each patient to consider their goals while also informing them about therapy limitations and associated risks, ultimately resulting in an increased perception of quality of life. Patients with genetic disorders require special care and tailored surveillance. As our patient's report described, new symptoms or findings may be related to factors such as penetrance, type, and mutation location. It is essential to perform close follow-ups for patients to optimize prognosis and manage complications, particularly for diagnoses without a definitive cure. All efforts towards clear communication should be pursued to ensure that patients understand their disease, natural course, and surveillance.

This case report describes a concurrent occurrence between FAP, GS, and achalasia not previously described in the current literature.

CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

ETHICS STATEMENT

Informed consent was obtained from the patient to publish the case details.

ORCID

Alejandro Llanos-Chea https://orcid.org/0000-0002-8882-2900

REFERENCES

1. Vasen HFA, Tomlinson I, Castells A. Clinical management of hereditary colorectal cancer syndromes. *Nat Rev Gastroenterol Hepatol*. 2015;12(2):88-97.
2. Dinarvand P, Davaro EP, Doan JV, et al. Familial adenomatous polyposis syndrome: an update and review of extraintestinal manifestations. *Arch Pathol Lab Med*. 2019;143(11):1382-1398.
3. Schlottmann F, Herbella F, Allaix ME, Patti MG. Corrigendum to: "Modern management of esophageal achalasia: from pathophysiology to treatment," Curr Prob Surg 2018;55(1):1-38. *Curr Probl Surg*. 2019;56(2):91.
4. Kahrilas PJ, Bredenoord AJ, Fox M, et al. The Chicago classification of esophageal motility disorders, v3.0. *Neurogastroenterol Motil*. 2015;27:160-174.

5. Talseth-Palmer BA. The genetic basis of colonic adenomatous polyposis syndromes. *Hereditary Cancer Clin Pract*. 2017;15:5.
6. Gupta P. Primary versus secondary achalasia: a diagnostic conundrum. *Trop Gastroenterol*. 2015;36(2):86-95.
7. Richter JE. Tailoring therapy for achalasia. *Gastroenterol Hepatol (N Y)*. 2020;16(5):249-257.
8. Gillies CL, Farrukh A, Abrams KR, Mayberry JF. Risk of esophageal cancer in achalasia cardia: a meta-analysis. *JGH Open*. 2019;3(3):196-200.

How to cite this article: Lorusso Vivas GA, Brady A-C, Llanos-Chea A. Familial polyposis syndrome and achalasia in a young adult. *JPGN Rep*. 2025;6:166-169.
doi:10.1002/jpr3.12161