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Case Report
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Attenuated familial adenomatous polyposis presenting as an abdominal wall mass

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ABSTRACT

A case is discussed involving a 25-year-old female with attenuated familial adenomatous polyposis that presented as an abdominal wall mass. The patient eventually underwent a restorative proctocolectomy with ileal pouch anal anastomosis.

Key words: *Familial adenomatous polyposis, restorative proctocolectomy*

Introduction

Familial adenomatous polyposis (FAP) is a dominantly inherited genetic cancer syndrome caused by inactivation of a tumor suppressor gene. Patients with the disease develop 100-1000 polyps in the gastrointestinal tract. Attenuated FAP is a part of the spectrum of the disease in which there are only 60-100 polyps in the colon [1,2].

Often presenting with gastrointestinal bleeding, diarrhea, obstruction, or polyp prolapse, we did not find a report of FAP presenting as an abdominal wall mass in the literature [1-3]. This renders the case covered here of interest.

Case Report

A 25-year-old female from a coastal town in the Philippines presented with a right lower quadrant (RLQ) mass.

She initially complained of intermittent bilious vomiting, anorexia, and generalized colicky abdominal pain and was managed as a case of gastrointestinal tuberculosis (GITB) based on computed tomography (CT) scan findings alone.

At the time of the patient's consult, she presented with a 6 cm fungating, friable mass at the RLQ with foul-smelling mucoid discharge. Digital rectal examination (DRE) revealed multiple sessile masses occupying almost the entire circumference of the rectum, the most distal extent reaching 3 cm from the anal verge (FAV).

An incision biopsy of the abdominal wall mass showed an adenomatous polyp. The patient was prepared for surgery and eventually underwent a restorative proctocolectomy (RPC) with ileal pouch (S-pouch) anal anastomosis (IPAA). A loop ileostomy

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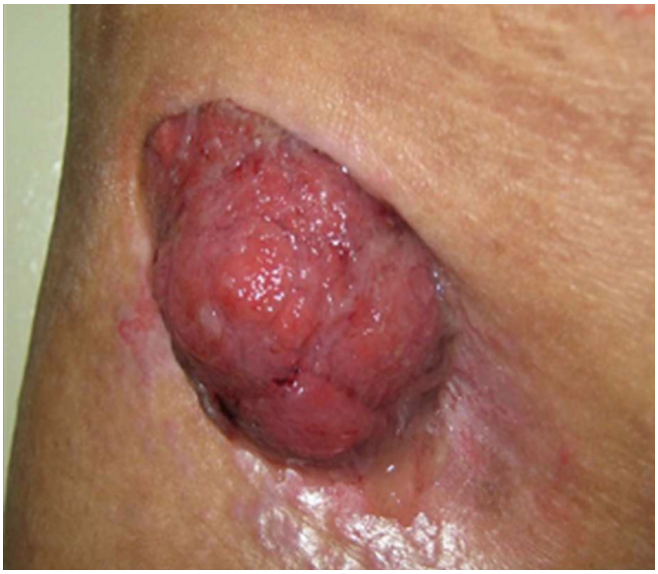

Figure 1. A 6 cm right lower quadrant mass in a 25-year-old female with mucoid discharge.

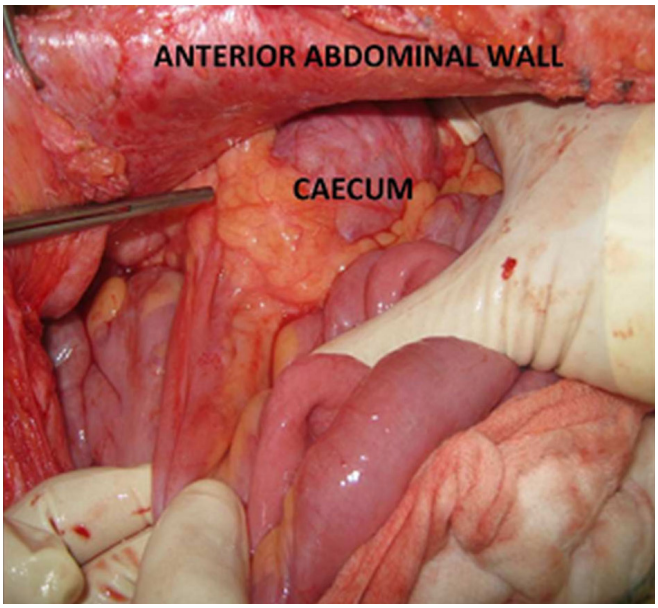

Figure 2. The cecum with polyps protruding through the anterior abdominal wall.

was exteriorized for proximal fecal diversion.

The patient was sent home on the 5th day post-operatively. The final histopathology report was multiple tubular adenomatous polyps.

Discussion

Familial adenomatous polyposis (FAP) is a dominant autosomal inherited disorder where patients are expected to develop colorectal cancer by the age of 40. Surgical prophylaxis, therefore, is recommended for patients diagnosed with FAP. The choice of surgical procedure is dictated by the number of polyps involved, the location of the polyps, and the presence of histologically-proven malignancy [1,2].

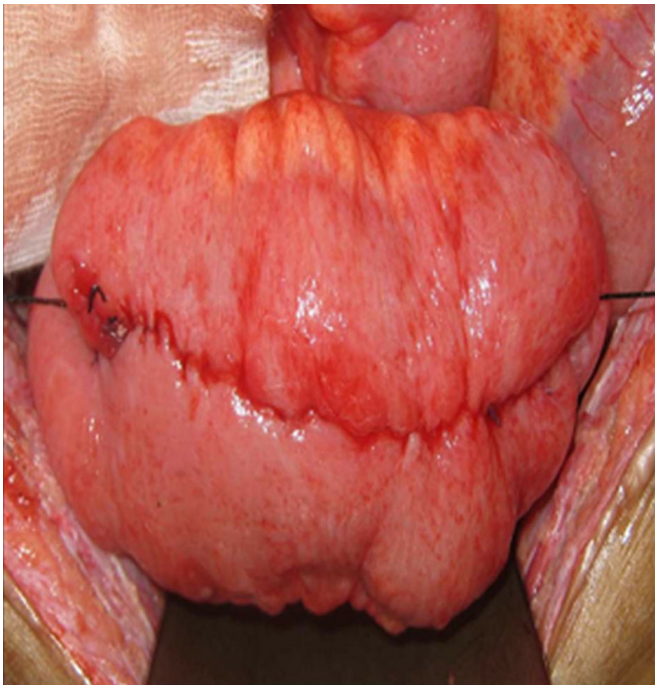

Figure 3. The constructed S-pouch during the restorative proctocolectomy.

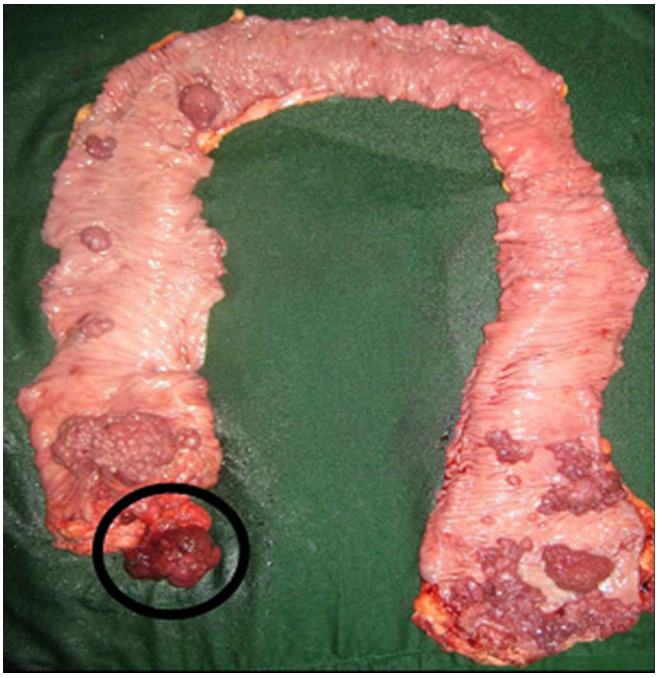

Figure 4. Opened specimen of the colon and rectum showing multiple polyps. Encircled is the mass that was protruding from the anterior abdominal wall. UP-PGH, 2012.

FAP often presents with symptoms of gastrointestinal bleeding, obstruction, weight loss, or prolapse of a polyp through the rectum. Extraintestinal manifestations, such as osteomas, central nervous tumors, congenital hypertrophic retinal pigment epithelium, and thyroid cancer, are often observed [3].

The aforementioned case is unusual in that FAP presented as an abdominal wall mass. Initially, the gross

appearance of the abdominal wall mass coupled with its having violated normal anatomic planes aroused suspicion of a possible malignant entity. Histopathology, however, confirmed this to be benign. The patient was, thus, scheduled for an RPC with IPAA.

In the Philippines, it is unfortunate that genetic counseling and screening for FAP and other hereditary conditions are not widely practiced. Often, patients are seen at late stages where malignant transformation has already occurred.

Conclusion

Being presented with this case, medical practitioners should be made aware of the possibility of unusual signs and symptoms relating to FAP. An early referral to a specialist is warranted to ensure prompt intervention. This further highlights the need for a more aggressive approach towards early detection of FAP and similar hereditary colorectal conditions in the Philippines.

Conflict of interest statement

The authors have no conflicts of interest to declare.

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