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IMAGE | COLON

A Patient With Gardner's Syndrome and Familial Adenomatous Polyposis Presenting With Extra-abdominal Desmoid Tumors and Diffuse Intestinal Polyposis

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Case Report

A 26-year-old Haitian-American man, diagnosed with a desmoid tumor of the right foot 2 years earlier, presented with pain and purulent bloody discharge from an ulcerated right foot mass (Figure 1). His physical exam revealed dental abnormalities including the loss of teeth. Radiographic imaging studies revealed a multi-lobulated right foot mass that measured over 9 x 8 cm and multiple subcutaneous cystic masses on his back, right upper leg, and head, measuring up to 2.4 cm. Colonoscopy and upper endoscopy performed due to suspicion for Gardner's syndrome demonstrated more than 50 polyps throughout the colon (Figure 2), more than 25 polyps in the duodenum (Figure 3), and innumerable polyps in the fundus and body of the stomach. Biopsies of the colonic and duodenal polyps were consistent with tubular adenomas. An ophthalmologic exam did not reveal congenital hypertrophy of the retinal pigment epithelium. Adenomatous polyposis coli (APC) gene sequencing revealed that the patient was heterozygous for a deleterious mutation of the APC gene. The patient subsequently underwent a Whipple procedure and a total proctocolectomy with ileo-anal J-pouch.

Familial adenomatous polyposis (FAP) is a rare condition that occurs in 1/10,000 to 1/30,000 newborns, and is responsible for approximately 1% of colorectal carcinomas in the world. Gardner's syndrome, a subtype of FAP, is characterized

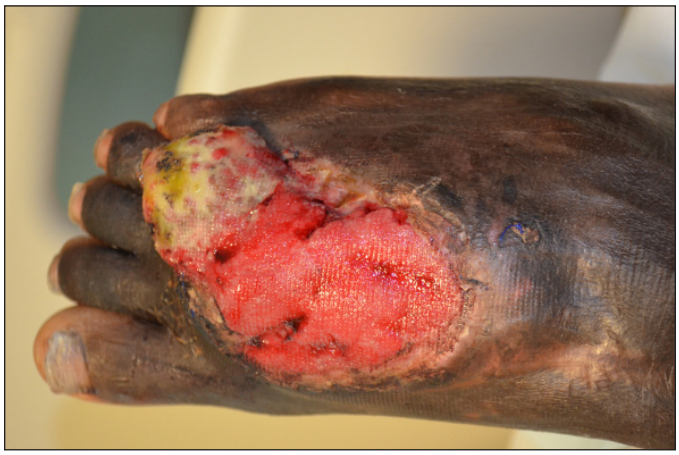

Figure 1. Desmoid tumor on the right foot.

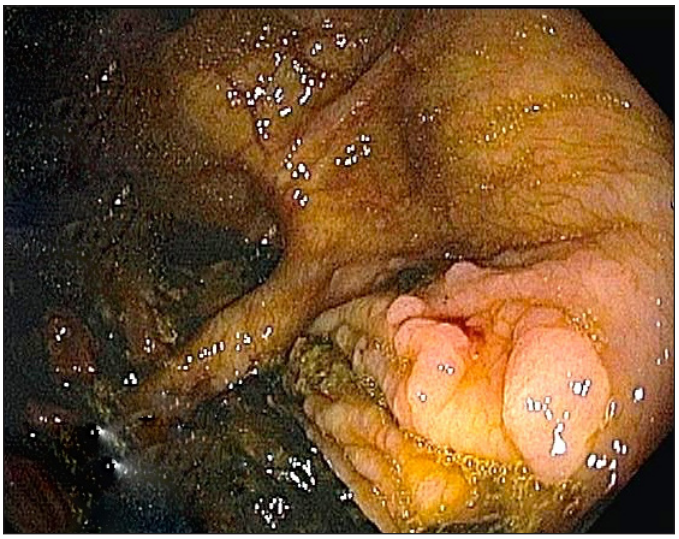

Figure 2. Endoscopic view of colon polyps.

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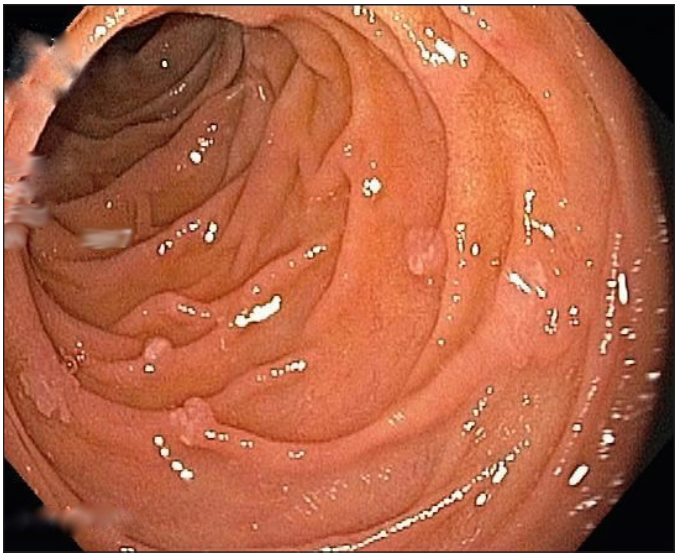

Figure 3. Endoscopic view of duodenal polyps.

by multiple adenomatous polyps in the colon, along with extracolonic findings including desmoid tumors, osteomas, lipomas, dental abnormalities, epidermoid cysts, and duodenal adenomas.[1,2] The pathogenesis behind this condition involves an autosomal dominant loss of function mutation in the tumor suppressor gene APC, located on chromosome 5q21-q22.[3] Although the patient was diagnosed with an extraintestinal desmoid tumor 2 years earlier, the association with Gardner's syndrome was not initially recognized. Most desmoid tumors occur intra-abdominally or on the abdominal wall in 10–20% of patients with Gardner's syndrome, with a peak incidence at 30 years of age. Extra-abdominal desmoid tumor occurs in fewer than 10% of patients.[2]

Disclosures

Author contributions: HD Patel wrote the manuscript and assisted in the literature review. BA Schwartz wrote, edited, and proofread the manuscript. MZ Rahman organized and facilitated the literature review and discussion. EB Grossman assisted with manuscript writing and final approval, and is the article guarantor.

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References

1. Wennstrom J, Pierce ER, McKusick VA. Hereditary benign and malignant lesions of the large bowel. *Cancer*. 1974;34(3)(suppl):850–7.
2. Groen EJ, Roos A, Muntinghe FL, et al. Extra-intestinal manifestations of familial adenomatous polyposis. *Ann Surg Oncol*. 2008;15(9):2439–50.
3. Lamlum H, Ilyas M, Rowan A, et al. The type of somatic mutation at APC in familial adenomatous polyposis is determined by the site of the germline mutation: A new facet to Knudson's "two-hit" hypothesis. *Nat Med*. 1999;5(9):1071–5.