Purpose: Case: A 71-year-old female underwent a screening colonoscopy at the age of 27 due to the early death of a parent from colon cancer. At the time, she was asymptomatic with no significant PMHx. Colonoscopy findings included a plethora of polyps concerning for familial adenomatous polyposis (FAP) by clinical criteria. Subsequently, the patient underwent a colectomy with end ileostomy. Screening EGD at that time revealed a Spigelman score correlating with stage 0 disease. She was initiated on a malignancy-screening protocol and averaged a screening endoscopy and ileoscopy every 5 years. All procedures were normal until an EGD at age 52 revealed malignant duodenal adenomatosis. She underwent a pylorus-sparing Whipple procedure. Future ileoscopy revealed a tubular adenoma that was removed without incident at age 65. At age 70, an EGD revealed flat low-grade dysplasia in the antrum with intestinal metaplasia. Repeat EGD 6 months later showed a 2 cm flat polyp in the efferent limb and a 3 cm flat polyp past the pylorus. Pathology revealed tubular adenomas with focal high-grade dysplasia and carcinoma. Functionally, the patient had greatly deteriorated by this point. Because of the patient’s poor general health and the nature of the current lesions, endoscopic attempt at removal was considered futile. Surgical evaluation concurred that the patient was not an ideal candidate for surgical intervention. Consequently, after extensive discussion with the patient, the decision was made to proceed with a palliative care approach.

Discussion: Our case depicts the interesting scenario of a patient with FAP who, despite appropriate screening and an extensive surgical history, still ultimately suffered from a malignancy associated with her diagnosis. As an inherited colorectal cancer syndrome, FAP accounts for 1% of colorectal cancer. More than 80% of patients will form polyps in the stomach and small intestine. This has prompted screening guidelines by both the American and European societies (with surveillance intervals based on the calculated Spigelman score). The screening employed in our case is consistent with the recommendations of the American guidelines, and yet our patient was still ultimately diagnosed with an incurable malignancy as a result of her FAP. This case illustrates the importance of recognizing the diagnosis of FAP in patients early and appropriately. Furthermore, as the aforementioned recommendations are from 2006 and 2008 respectively, an updated population review of patients suffering from FAP from the last several years should be considered. This could potentially lead to more stringent recommendations and the ability to further decrease preventable malignancies in this population.

Methods: N/A
Results: N/A
Conclusion: N/A

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Purpose: To show the efficacy of chromoendoscopy in detecting polyps not seen by white light sigmoidoscopy in a patient with familial adenomatous polyposis (FAP) who underwent an ileo-rectal anastomosis and had a remaining small cuff of rectal tissue.

Introduction: Chromoendoscopy enhances the detection of neoplasia in the colon and rectum. Patients with neoplastic polyps, particularly those with multiple polyps, are at increased risk of developing colorectal cancer. These lesions presumably would be missed with conventional colonoscopy and could contribute to the interval cancer numbers on any surveillance program.

Case report: A patient with FAP underwent an ileo-rectal anastomosis and had screening flexible sigmoidoscopy regularly for removal of adenomas. On her latest sigmoidoscopy after removal of all adenomas visible by white light, the cuff was sprayed with indigo carmine and additional adenomatous colon polyps were identified and removed (FIG).

Discussion: Chromoendoscopy vs. white light colonoscopy results in a statistically significant difference between the groups regarding the total number of adenomas detected and with significantly more diminutive (<4 mm) adenomas detected. 1 Importantly, in one study eight diminutive lesions had foci of high grade dysplasia; therefore, chromoendoscopy may especially benefit patients with a high risk of colorectal cancer, as in our patient.2 We found that chromoendoscopy enhanced the detection of adenomatous polyps in the remaining distal colon in this patient with FAP.

Conclusion: Chromoendoscopy using indigo carmine enhances the detection of adenomatous polyps in the colon cuff in patients with FAP and should be utilized in these patients at high risk for colon cancer.

Purpose: Muir-Torre syndrome (MTS) is a rare autosomal dominant familial cancer syndrome diagnosed with occurrence of cutaneous sebaceous tumors along with at least one visceral malignancy, most frequently gastrointestinal carcinoma. We report a case of a 46-year-old gentleman who was diagnosed with Stage II B colorectal cancer in 2002 treated with partial colonic resection. Surveillance colonoscopy in 2005 revealed tubular adenoma which was resected endoscopically. His father was diagnosed with colon cancer at the age of 42 and pancreatic cancer at the age of 62. Paternal uncle had colon cancer at the age of 56 and paternal grandmother had brain tumor at the age of 66 years indicating a possibility of Lynch syndrome (Amsterdam criteria II). Subsequently the patient underwent MLH1 and MSH2 sequencing and was found to have an MSH2 mutation known as 1003insA. All his siblings underwent MSH2 site specific testing with two siblings testing positive for familial MSH2 mutation. In 2013, the patient presented to the dermatologist for an ulcerating lesion on the nose. The biopsy of the lesion showed sebaceous carcinoma (Figure 1). The diagnosis of Muir-Torre syndrome was made based on history of colorectal cancer, MSH2 mutation, and development of sebaceous carcinoma.

MTS is a rare syndrome which is diagnosed in patients who are found to have sebaceous carcinoma with visceral malignancy. Sebaceous carcinoma is a highly aggressive tumor and should be suspected in Lynch syndrome patients with skin lesions. Early diagnosis and close surveillance for visceral malignancies is essential to achieve a better outcome.

Methods: N/A

Results: N/A

Conclusion: N/A
REVIEWER COMMENTS:
Sita Chokhavatia: [No Comments]
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