Clicking Time Bomb: Unusual Management Approach of Gardner's Syndrome

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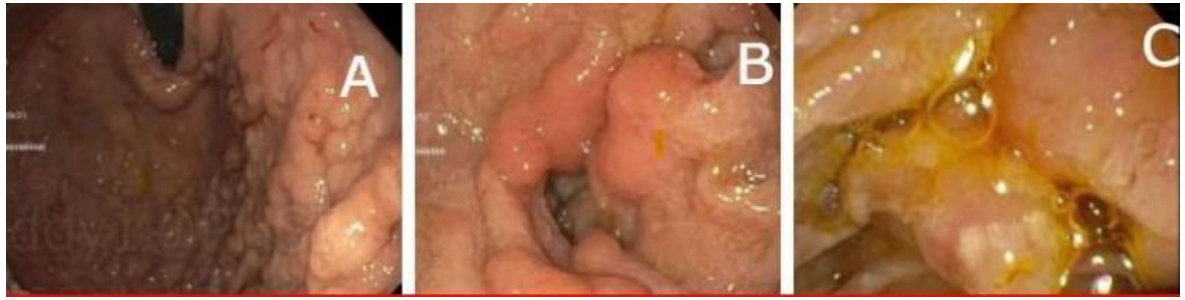
Introduction

Familial adenomatous polyposis is an autosomal dominant inherited disease due to mutation in chromosome 5q APC tumor suppressor gene (adenomatous polyposis coli)[i]. Numerous colonic polyps develop which progress to colonic cancer inevitably by the age of 35 in up to 95 %. Other Intestinal manifestations include gastric and duodenal polyps which can progress to malignancy. Extraintestinal manifestations include CHRPE (congenital hypertrophy of retinal pigment epithelium), CNS tumors, endocrine malignancy like thyroid papillary cancers, osteomas, skin and soft tissue tumors like fibromas, sebaceous cysts, and desmoid tumors. A prophylactic proctocolectomy is practiced against colon cancer in FAP patients leaving them with reduced length of intestines. Patients experience significant morbidity and dinical picture becomes further complicated if further polyps develop in stomach or small intestine.[ii] Here in this case, we report a case of FAP with previous proctocolectomy presenting with gastric and duodenal adenomas.

Case Presentation

A thirty-six-year-old female with history of familial adenomatous polyposis (FAP), diagnosed at the age of sixteen, and underwent proctocolectomy soon thereafter. She undergoes bi -annual endoscopies for surveillance. She also has a history of bilateral adrenal adenomas, desmoid tumor in the proximal lower extremities bilaterally, and Thyroid cancer – for which she underwent thyroidectomy and radiation. She presented to the emergency department complaining of generalized weakness associated with headache and exertional dyspnea. She denies any other complaints. Patient was afebrile, heart rate 75 beats/minute, 16 breaths/minute, saturating 94% on room air. On the physical exam, she exhibited conjunctival pallor and capillary refill time of more than 3 seconds. Hemoglobin was 6.1, mean corpuscular volume 52.6, red cell distribution width 26.7, iron level 22, ferritin level 3, iron saturation 4.1, and total iron binding capacity 535. The patient was admitted for symptomatic anemia. Hemoglobin improved to 8.6 after two units pure red blood cell transfusion. Subsequently, all symptoms resolved. She was discharged with outpatient follow up for endoscopy a week later, for surveillance. Endoscopy showed innumerable sessile polyps located diffusely in the proximal stomach with antral sparing as well as large sessile polyps in the duodenal bulb and first portion of duodenum.



A. Gastric Polyps, B/C Duodenal Polyps

Discussion

The incidence of gastric and duodenal polyps in a patient with FAP is 90%. The risk of progression to periampullary carcinoma is 3-12 % in the patients[iii]. However, they can also present with malabsorption and iron deficiency anemia. Duodenal polyps can be monitored and dassified as proposed by Spigelman which is based on polyps' size and number, histology and grade of dysplasia. It can be divided into 4 stages based on a scoring from 0-12[iv]. Patients with duodenal polyps can be managed by endoscopic surveillance every 1-5 years depending on the Spigelman staging. Celecoxib is suggested for stage 3. Endoscopy with EUS and duodenectomy (pancreas sparing or pylorus sparing) is recommended for stage 4.

Conclusions

Knowledge about Spigelman dassification is required in managing duodenal polyps in patients with FAP. Given the prophylactic proctocolectomy, every effort has to be made to preserve the rest of the intestine in these patients.

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References

i. Kinzler KW, Nilbert MC, Su LK, Vogelstein B, Bryan TM, Levy DB, Smith KJ, Preisinger AC, Hedge P, McKechnie D, et al. Identification of FAP locus genes from chromosome 5q21. Science. 1991 Aug 9;253(5020):661-5. doi: 10.1126/science.1651562. PMID: 1651562
ii. Half E, Bercovich D, Rozen P. Familial adenomatous polyposis. *Orphanet J Rare Dis*. 2009;4:22. Published 2009 Oct 12. doi:10.1186/1750-1172-4-22
iii. inha J, Williamson RC. Villous adenomas and carcinoma of the duodenum in Gardner's syndrome. *Postgrad Med J*. 1988;64(757):899-902. doi:10.1136/pgmj.64.757.899
iv. Groves CJ, Saunders BP, Spigelman AD, Phillips RK. Duodenal cancer in patients with familial adenomatous polyposis (FAP): results of a 10 year prospective study. *Gut*. 2002;50(5):636-641. doi:10.1136/gut.50.5.636