A Rare Case of Synchronous Familial Adenomatous Polyposis and Endometrial Carcinoma

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INTRODUCTION

- Familial adenomatous polyposis (FAP) is a rare autosomal dominant disease characterized by numerous polyps in the colon.
- It is caused by a germline mutation of the adenomatous polyposis coli (APC) gene.
- FAP patients have guaranteed risk of developing colorectal cancer if untreated and increased risk for extra-intestinal malignancy.
- Endometrial malignancy is not known to be associated with FAP, here we present a rare case of synchronous FAP and endometrial carcinoma.

CASE SUMMARY

- A 51-year-old female with family history of autosomal-dominant-patterned colon cancer, subtotal colectomy at 17 due to multiple polyps, ileostomy with a J-pouch at 35, and recent upper endoscopy suspicious for ampullary adenoma, presented with one-month history of fatigue and night sweats.
- Review of system was positive for heartburn and easy bruising. The patient did not have formal genetic testing.
- She has close follow-ups with yearly surveillance upper GI endoscopy (EGD), flexible sigmoidoscopy, and thyroid ultrasound.
- At 44, she underwent dilation and curettage due to menorrhagia; samples revealed endometrial cancer, which led her to undergo a bilateral salpingo-oophorectomy.

FIGURES

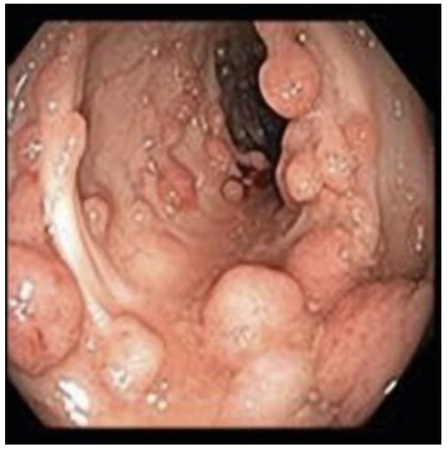

Figure 1. Colonoscopy showing many polyps in distal colon

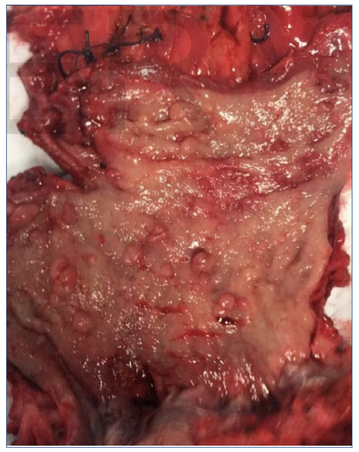

Figure 2. Post-colectomy specimen shows multiple polyps

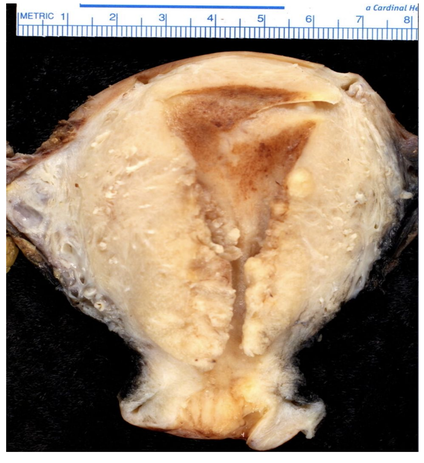

Figure 3. Gross hysterectomy specimen showing tumor growth in endometrium invading slightly more than half of myometrium in thickness

DISCUSSION

- Synchronous endometrial and ovarian cancers in FAP are rare.
- Another case reported a 57-year-old female FAP found to have bilateral ovarian microcystic stromal tumors (MCSTs) and endometrial carcinoma. Histopathology from the MCSTs and thyroid was both positive for beta-catenin, an important marker in FAP.
- MCST is a rare subtype of ovarian cancer found to be concurrent with FAP on several occasions.
- Despite not having genetic testing, the large number of polyps and autosomal dominant pattern of inheritance in our patient are consistent with FAP, as opposed to Lynch or MUTYH-associated polyposis syndromes.
- It is important to look at histopathology in our patient to see if there is any concurrent genetic expression with FAP
- This case implies there might be benefits in future screening of endometrial/ovarian cancer in patients with FAP.

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