

# Colorectal Cancer in a Pregnant Woman with Familial Adenomatous Polyposis

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## Citation

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## Abstract

Familial adenomatous polyposis is a disease inherited in an autosomal dominant fashion. Adenomatous polyps diffusely located in the colon and rectum characterize the disease. These polyps may also be identified in other parts of the GI tract. Afflicted patients usually develop primary colorectal cancer by their fourth decade of life. A mutation of the APC gene on chromosome 5q21 is responsible for FAP. This gene is a natural tumor suppressor and mutation allows the development of colorectal cancer. As with any colon cancer, metastasis to the liver is a possibility and occurs in 10%-20% of cases leaving patients with a median survival of 12-24 months. This report describes a 29-year-old female presenting with undiagnosed FAP and colorectal cancer. Her case was further complicated by a live intrauterine pregnancy and later to liver metastasis.

## CASE REPORT

A 29-year-old pregnant female presented to a community hospital emergency department complaining of sharp, lower abdominal pain with blood-tinged emesis and scant hematochezia. The patient stated that she had been suffering from 2 months of diarrhea followed by the last 2 weeks before presentation with constipation. She denied vaginal discharge or cramping and reported being approximately 7 weeks pregnant. She had significant family history of colon cancer with her father being diagnosed and requiring colectomy at 25 years of age. Her grandmother also had a history of colon polyps. Patient denied smoking, alcohol or drug abuse.

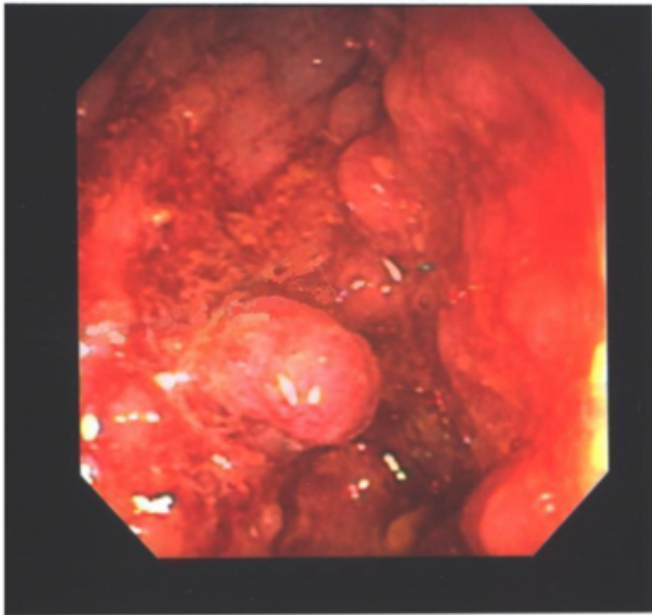
On exam, she was afebrile and in no acute distress. Her vital signs were stable. Neck, heart, lung, and extremity exams were unremarkable. Abdominal exam revealed mild obesity, guarding, and severe tenderness of the left lower quadrant without masses or organomegaly.

Labs and imaging studies revealed the patient had a microcytic/hypochromic anemia, low albumin level, and beta-HCG level consistent with a gestation of 1-2 months. An abdominal and pelvic ultrasound indicated only minimal wall thickening of the sigmoid colon, a live intrauterine pregnancy just less than 7 weeks, a small amount of free fluid in the pelvis, and a small right ovarian cyst measuring 2.5cm in diameter.

Due to her current pregnancy, it was determined the next best step would be colonoscopy. Upon exam, polyps were found extending from the mid-portion of the rectum distally to the 30cm mark of the sigmoid colon proximally. The colonoscope could not be advanced beyond 30cm due to a hard obstructing mass (figure 1). Multiple biopsies were taken at this location before withdrawing the colonoscope. Frozen section revealed a diagnosis of tubular adenoma with slight atypia and grossly, adenomatous polyposis coli was suspected.

**Figure 1**

Figure 1: Obstructing sigmoid tumor identified with colonoscopy



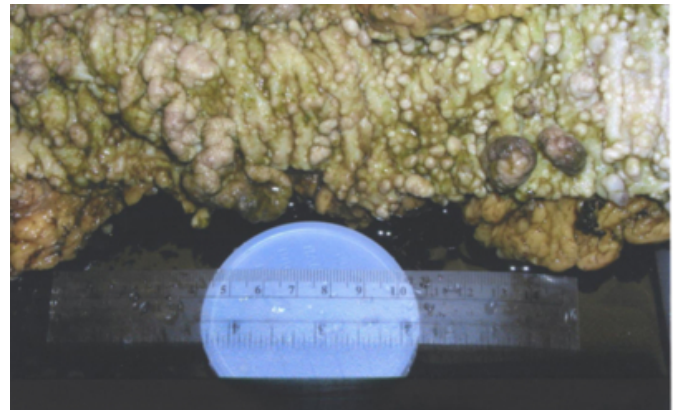
The results of the partial colonoscopy and treatment options were discussed with the patient and her family. The patient elected to have a total abdominal colectomy with ileorectal anastomosis and close observation of the remaining rectal stump. This was deemed the best option to avoid an ileostomy and extra stress on her pregnancy.

When surgery was performed the next day, a bulky sigmoid mass with local tumor reaction was encountered. Upon mobilization, there was pericolonic, intermesenteric abscess. There were no palpable liver masses or mesenteric adenopathy noted. Biopsies were taken from the sigmoid, descending, and ascending colon as well as the resected portion of the distal ileum. The pathology report read as follows:

- Descending and sigmoid colon – moderately differentiated adenocarcinoma extending through the muscularis mucosa into the pericolonic soft tissue (figure 2), abscess and granulation tissue surrounding the tumor, marked adenomatous polyposis coli, and metastatic carcinoma in 2 of 14 lymph nodes. Therefore, this tumor was staged as IIIB (T3N1Mx – metastasis workup not completed due to pregnancy) using the TNM classification of colorectal cancer.<sup>4</sup>
- Distal ileum and ascending colon – marked adenomatous polyposis coli.

**Figure 2**

Figure 2: Dissected sigmoid colon showing FAP

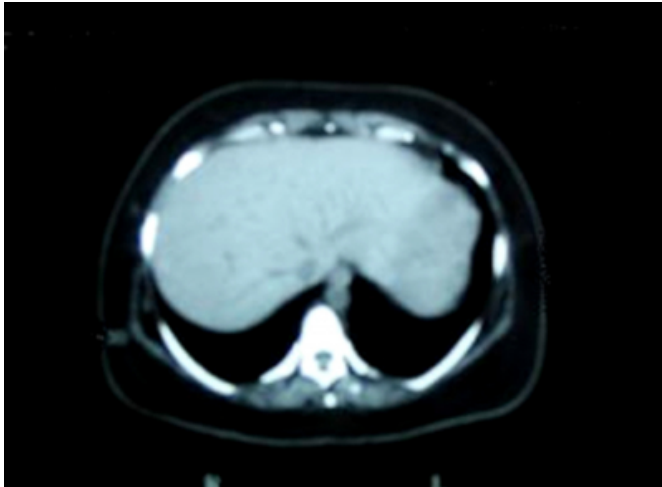


Upon resection of the colon and cancer diagnosis, an oncologist recommended the patient receive chemotherapy. At this time, the patient's pregnancy was only in the 8<sup>th</sup> week of gestation. Therefore, the patient elected to delay metastatic workup and chemotherapy until after her baby was born. She continued to be followed by an oncologist and surgeon and had a relatively normal pregnancy.

After giving birth to a healthy child, the patient began her metastatic workup for colon cancer. CT of the abdomen revealed a large mass in the left lobe of the liver (figure 3). Her CEA level at this time was 50ng/mL (normal reference range = 0 – 3.0 ng/mL). No other metastases were found. After reviewing potential options, the patient was optimistic and decided to have a left hepatic lobectomy. The surgery progressed without complications and a tumor measuring 11.2cm x 7.8cm was resected, and a cholecystectomy was also performed. Frozen section of the resected tumor showed metastatic adenocarcinoma consistent with primary colonic cancer. The patient recovered well from surgery and was discharged on the sixth post-op day. A CEA level evaluated one-week post-op had decreased to 4.8ng/mL.

**Figure 3**

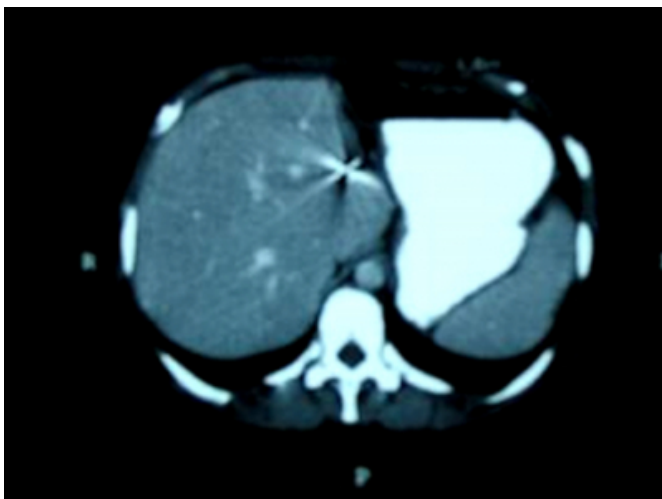
Figure 3: CT scan of the liver with evidence of left lobe metastasis



A few weeks later, the patient began a chemotherapy regimen often used for colorectal cancer known as FOLFOX4 consisting of Oxaliplatin, 5-Fluorouracil, and Leucovorin dosed every 2 weeks for ten cycles. She continued to follow-up as needed. After finishing her course of chemotherapy, CT scan (with contrast) of the thorax, abdomen, and pelvis revealed no evidence of metastasis (figure 4). Proctoscopy of the rectal stump was also performed at this time and was negative for polyps.

**Figure 4**

Figure 4: Follow-up CT scan after left hepatic lobectomy



## DISCUSSION

Colon cancer in patients with FAP is very well documented in the literature. As previously mentioned, it is an autosomal dominant disease with very high penetrance approaching 100%. The current incidence of FAP is estimated at 1 in

8000 people with this number being maintained by cases arising through de novo APC gene mutation. Mutations in the APC gene usually result in missense or frameshift of the genetic code consequently producing abnormal truncated protein products.<sup>1</sup> These abnormal proteins alter the functioning of the cell cycle, differentiation, and possibly apoptosis.<sup>5</sup>

Familial adenomatous polyposis is diagnosed most commonly now through colorectal screening. When a person is identified with the disease, family members should be tested to see if they also have the APC gene mutation. If tested positive, the family member must receive routine colonoscopy to treat any existing polyps. They should also be definitively treated with total abdominal colectomy and ileorectal anastomosis on an elective basis. Further follow-up every 3-6 months with proctosigmoidoscopy of the remaining rectal stump is advised for the remainder of the patient's life. A total proctocolectomy could also be performed to remove all colorectal tissue, but a permanent ileostomy is often required.<sup>6</sup> The patient and his or her family should be educated about the disease so they can choose the best surgical treatment to accommodate the patient's lifestyle.

Those patients similar to our case subject, who present and are diagnosed with FAP after severe symptoms, have increased chance of advanced colorectal cancer and metastasis. Statistically, 10% - 20% of colorectal cancers will metastasize to the liver as a result of portal venous drainage of the bowel, and those patients have a median survival of only 12-24 months. Clearly, if the liver mass is resectable, the prognosis improves somewhat and 5-year survival is estimated at 26%-40%. However, 75% of patients with resectable liver masses have a metastatic recurrence within two years.<sup>7</sup> Adjuvant chemotherapy may then be employed to increase total time of disease-free survival.

## CONCLUSION

This report should emphasize the necessity of surgical treatment for FAP along with prompt metastatic workup and chemotherapy for colorectal cancer. With regards to our case subject, the lack of appropriate counseling to her family placed the patient at increased risk. Likely, she was not educated of the necessity for frequent colon exam or prophylactic colectomy as an adolescent. In addition, socioeconomic considerations potentially explain why the patient waited so long to seek medical attention. Fortunately, the patient had a relatively normal pregnancy and delivered a

healthy infant. Currently, she is doing well and maintains disease-free survival one-year after resection of the liver metastasis. Since it is estimated that the incidence of colorectal cancer during pregnancy is 0.002%, and of those only 32 cases have been reported above the peritoneal reflection<sup>8</sup>, this is certainly is a rare case worth mention.

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