Total Colectomy with Ileorectal Anastomoses in a Patient with Asymptomatic and Genetically Negative Polyposis Syndrome

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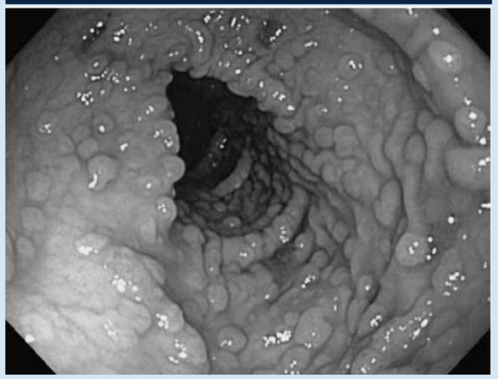
Introduction

- Familial adenomatous polyposis (FAP) is a condition in which more than 100 polyps line the colon as seen in Figure 1.[1]
- Anywhere from 1 in 8,000 to 35,000 people are diagnosed with FAP.[1-3]
- The majority of patient diagnosed with FAP are diagnosed between ages of 20 to 40 years old.[2]
- FAP is an autosomal dominant condition. There is a 50% chance of an individual having FAP if there is a first degree relative with FAP.[4]
- People with FAP have nearly a 100% chance of developing colorectal cancer (CRC).[1]
- FAP makes up less than 1% of CRCs.[3-5]
- 70% of CRCs develop from adenomatous polyps.[6]
- CRC is the 3rd leading cause of death for men and women.[5]
- Signs and symptoms warranting screening for CRC or polyps that are seen in FAP patients include:[2]
  - Positive fecal occult
  - Constipation
  - Diarrhea
  - Bloating
  - Unintentional weight loss
  - Abdominal pain
  - Anemia
- CRC screening recommendations as seen in Figure 2.
- FAP is known to be caused by genetic mutations in the adenomatous polyposis coli (APC) gene or Human MutY homolog (MUTYH) gene .[1]
- Mosaicism is when mutations are only present in a proportion of cells in the body. These are epigenetic mutations occurring postzygotically.[4]
- FAP treatment is with colectomy which is also prophylactic against CRC.[1,7-9]
- People with factors such as male sex, obesity, longer operation time, and irritable bowel disease are at increased risk for complications following colectomy.[8]
- Obesity specifically increases risk for longer procedure time, greater blood loss, need for laparotomy opposed to laparoscopy, hernia, leak, and longer hospital stay.[8]

FIGURE 1: Familial Adenomatous Polyposis during Colonoscopy[1]



Case Description

HPI:
53-year-old male presented for surgical consult following asymptomatic polyposis found on routine colonoscopy. Patient denied abdominal pain, bleeding from rectum, dark or bloody stools, diarrhea, constipation, or weight loss. He denied any family history of colon polyps or colon cancers.

Past Medical History:
- GERD
- Panic attacks
- Seasonal allergies

Medications:
- fexofenadine 180mg PO daily prn

Past surgical hx:
- Umbilical hernioplasty 2006
- Lipoma removal 1976
- Tonsillectomy
- Rhinoplasty

Allergies: No known drug allergies.

Family Hx: No history of colon cancer or colon polyps. No other significant family history.

Social hx:
- Never smoker
- No alcohol use
- No substance use

Review of systems: Unremarkable.

Vital signs:
- Blood pressure: 128/87mmHg
- Heart Rate: 85bpm
- Respiratory Rate: 20 breaths per min
- Temperature: 98.5F
- Oxygen saturation: 97% on room air
- BMI: 30.1 kg/m2

Physical Exam:
- Mouth: Mucous membranes moist.
- Eyes: EOM intact, conjunctiva pink, PERRLA.
- Cardiovascular: Normal rate and regular rhythm. Pulses 2+ bilaterally.
- Pulm: No wheezes, rales, or rhonchi.
- Abdominal: Abdomen is flat, no distention, active bowel sounds, soft, non-tender, no guarding, rebound, and no masses.
- Skin: Warm, dry. Capillary refill less than 2 seconds.
- Noncontributory musculoskeletal, neurologic, and psychiatric findings.

Diagnostic Tests:
Genetic tests:
- Negative APC gene mutation
- Negative MUTYH gene mutation

Colonoscopy:
- Several non-bleeding diverticula in sigmoid.
- Too numerous to count small sessile polyps scattered about the entire colon: cecum, ascending colon, and proximal transverse colon "mucosa was nearly carpeted with polyps".

Pathology Report from Colonoscopy:
- Tubular adenomatous polyps of cecum, ascending colon, transverse colon, and sigmoid colon.
- One non-adenomatous polyp demonstrated colonic mucosa with mild hyperplastic glandular changes.
- One tubular adenoma with prolapse type changes.
- Margins negative for adenomatous glands.

DDx: FAP, attenuated FAP, peutz-jegher syndrome, Lynch syndrome, hereditary mixed polyposis syndrome
Final Dx: Idiopathic polyposis of the colon

FIGURE 2: CRC Screening Recommendations[6]:

First degree relative with CRC or adenomatous polyps:
Colonoscopy at age 40 or 10 years before age of affected relative, whichever is first.
Interval colonoscopy every 5 years.

Average Risk individuals:
Fecal Immunochemical Test (FIT) every 1 year *or*
Colonoscopy every 10 years starting at age 45.

Unable or unwilling to undergo colonoscopy or FIT:
Multitarget stool DNA test every 3 years.
Flexible sigmoidoscopy every 5-10 years.
CT colonography every 5 years.
Colon capsule every 5 years.
Screening beyond 75 years of age is individualized.

FIGURE 3: Hospital Course

- **7/22**: Routine colonoscopy
- **7/29/22**: Hospital Admission &Total Colectomy
- **Colectomy Pathology Report**: 23 benign lymph nodes; Numerous tubular adenomas; Margins negative for adenoma; Negative for carcinoma
- **Post-op Day 5**: Post-surgical ROS positive for diffuse abdominal pain, diarrhea, nausea, vomiting; Diagnosed ileus; Made NPO; D/c antidiarrheal medications
- **Post-op Day 7**: NGT placed with 1.5L of fluid aspirated into suction canister; TPN began
- **Post-op Day 10-18**: Patient diet advanced as tolerated; Strict I&O's to ensure proper oral and IVF repletion
- **Post-op Day 18**: Discharged & Outpatient follow up scheduled

Patient Management and Outcome

Management:
Surgical: Laparoscopic converted open total abdominal colectomy with ileorectal anastomosis.

Outcome:
Successful total colectomy with ileorectal anastomoses with post-op complication of post-op ileus which was reversed. Patient tolerated diet well and maintained proper fluid intake and was discharged home. Patient followed up in outpatient clinic two weeks later.

Discussion

- Diffuse polyposis without findings of the associated genetic mutations for hereditary diseases that cause this presentation supports the concept of genetic mosaicism.[4]
- Genetic mosaicism is a potentially overlooked cause of de novo FAP and CRC. If it becomes an accepted etiology, it can be used to guide screening and diagnosis of FAP and CRCs. [4]
- CRC screening should be encouraged by primary care providers from ages 45-49 in average risked individuals to reduce incidence of advanced adenoma and CRC.[6] Earlier detection in younger patients can lead to better outcomes. [2]
- Colectomy is the only definitive cure for FAP, but other options should be explored due to younger patient's postponing or refusing surgery. Intensive endoscopic downstaging of polyp burden (IDP) may have a role in preventing CRC in mild to moderate FAP. [9]

Conclusion

- Genetic counseling should be considered and offered to patients and their families when diagnosed with conditions like FAP.
- Healthcare providers should ensure patients are screened at the proper age to discover polyps and reduce incidence of CRC morbidity and mortality.
- New research is showing mosaicism's role in hereditary conditions such as FAP and CRCs that should be incorporated into screening and diagnosis.
- More treatment options should be explored for FAP for patients that are at higher risk of complications with colectomy and those that refuse colectomy. IDP may provide one option for those unwilling to undergo colectomy.

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