A Rare Case of Stage III Colorectal Adenocarcinoma in a Child

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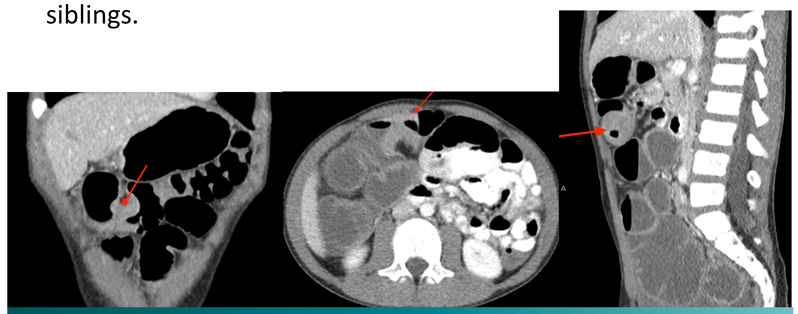
Introduction

- Colorectal carcinoma (CRC) in childhood is often sporadic, with approximately 10% to 30% of cases associated with a predisposing condition.[1]
- The presenting symptoms are typically nonspecific and coupled with the rarity of the diagnosis and low clinical suspicion, often results in delayed diagnosis, which leads to a significantly worse prognosis compared to the adult patient population, with one study reporting a 3-year recurrence free survival rate of 32% for pediatric patients, compared with 78% for adults.[2]
- Predisposing factors include familial adenomatous polyposis (FAP), juvenile polyposis syndrome (JPS), Lynch Syndrome, Gardner syndrome, Peutz-Jeghers syndrome, Turcot syndrome, and ulcerative colitis.[1,3]

Case

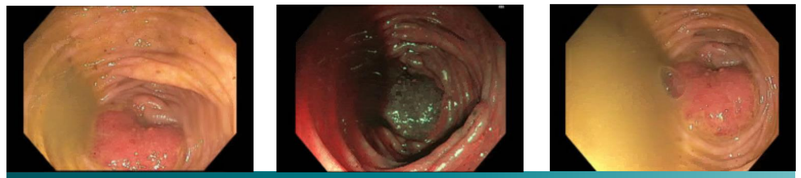
- An 11-year-old Hispanic male presented with concerns for bowel obstruction with history of increasing abdominal pain, emesis, weight loss and intermittent diarrhea for the past one year.
- Associated symptoms included poor appetite, unquantifiable weight loss, and new onset hematochezia. His medical history was significant for perforated appendicitis at 5 years of age.
- CT abdomen/pelvis with contrast was concerning for bowel obstruction with two separate areas of wall thickening and narrowing at the terminal ileum and within the proximal transverse colon creating an “apple core lesion.”
- Maternal family history was negative for tuberculosis, colonic polyps, and gastrointestinal tract cancers; paternal history was unavailable. The patient was noted to have two healthy older siblings.



Primary colonic mass of the transverse colon (red arrows) shown in coronal, axial, and sagittal planes on CT

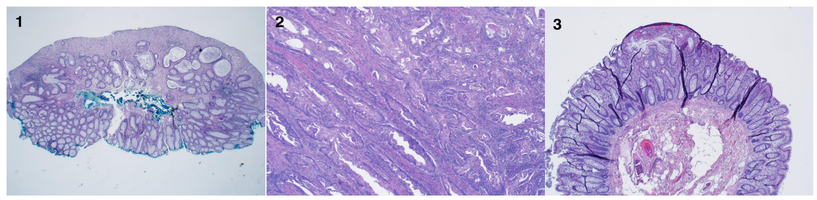
Case (Continued)

- Colonoscopy showed 4 polyps, all were < 5mm (2 in the rectum and 2 in the descending colon).
- A large circumferential mass was found partially obstructing the transverse colon, preventing further passage of the colonoscope.



Large circumferential mass partially obstructing the transverse colon as seen during colonoscopy

- 2 rectal polyps showed features consistent with juvenile-type hamartomatous polyps on pathology.
- Biopsies from transverse colon mass showed intramucosal adenocarcinoma.
- No polyps seen on an upper endoscopy.
- The patient underwent a total colectomy with end-to-end ileorectal anastomosis. The resected specimen demonstrated T4a N2a colorectal adenocarcinoma of the transverse colon with 4/83 lymph nodes positive for metastasis. 5 additional juvenile-type hamartomatous polyps were found throughout the colon.



1. Juvenile Polyp: surface erosion/inflammation with underlying dilated disorganized glands containing inspissated mucin, features consistent with a juvenile polyp, but not pathognomonic of this diagnosis as inflammatory polyps have similar features.
2. Adenocarcinoma: of the type demonstrating invasive glands through the muscularis propria.
3. Additional juvenile polyp found post-colectomy: shows surface inflammation/erosion, disorganized and branching underlying glands. The characteristic dilated glands with inspissated mucin are lacking given its small size. No features of colitis in adjacent tissue, excludes an inflammatory polyp.

Discussion

- CRC is a very rare malignancy in the pediatric population with a reported annual incidence of 1 case per 10 million adolescents less than 20 years old.[4]
- Comprises 1% of all pediatric neoplasms and despite being rare, CRC is the most common primary solid neoplasm of the GI tract in children.[5]
- While CRC can be associated with a variety of predisposing conditions, the majority of cases, approximately 90%[3], are sporadic.
- Upon screening the siblings, the patient's brother had 4 polyps, consistent with juvenile-type hamartomatous polyps, and the sister had 1 polyp.
- JPS is highly suspected in our patient as he met the criteria of > 5 juvenile-type polyps of the colon or rectum (our patient had 9); other criteria includes multiple juvenile polyps throughout the GI tract *or* any number of juvenile polyps and a positive family history of JPS.
- Molecular analyses of the KRAS, BRAF, NRAS, and PIK3CA mutations and for microsatellite instability were negative. A multi-gene panel for colorectal cancer with sequencing and deletion/duplication analysis of 25 genes is pending and includes those for JPS (*SMAD4, BMPR1A*).
- Screening first degree relatives for JPS is recommended as follows:
  - Screening colonoscopy at 12 to 15 years of age if asymptomatic and no known gene mutation detected.
  - If a mutation is known, the family member should undergo pre-symptomatic genetic testing, as well as genetic counseling and colonoscopy.[6]

Conclusion

- While CRC is an exceptionally rare diagnosis in a child, it should be on the differential for children presenting with prolonged abdominal pain of unclear etiology in the interest of early recognition and improved prognostic outcomes.
- Given the often sporadic nature of CRC in the pediatric population, this diagnosis should not be dismissed solely based on lack of family history, thus increased awareness among pediatricians and surgeons alike is necessary.

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