Pediatric Colorectal Cancer: Case Report

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ABSTRACT

**Background:** Colorectal cancer (CRC) predominately affects adults over the age of 50, however it occasionally occurs in young patients.

**Case:** This case observes the presentation and management of a 14-year-old African American male (AAM) who presented to the emergency department with a two-week history of insidious onset RLQ abdominal pain. Evaluation of suspected small bowel obstruction with computed tomography raised alarm for a mass lesion causing the obstruction. Subsequent barium enema, colonoscopy, and histological analysis diagnosed colorectal cancer. The patient then underwent surgery, chemotherapy, and radiation for stage IIIC CRC.

**Conclusion:** We use this case to argue for the development of a genetic panel that can screen for high-risk mutations so detected patients can pursue early and frequent screening protocols such as colonoscopy. We demonstrate the importance of early intervention by discussing the prognosis of patients that are discovered after a change in bowel habits.

**Keywords:** pediatric; colorectal cancer; case reports; genetics; oncology.
**Introduction:**
CRC is the second leading cause of cancer related death in the US overall. The prevalence of CRC reaches .4%, however the peak incidence occurs within the age range of 65 to 74. Most cases of non-hereditary colorectal cancer arise from the chromosomal instability pathway, where sequential mutations in oncogenes [e.g., APC, KRAS, TP53, DCC], cause development of an adenomatous polyp and progression of a polyp to cancer. Approximately 35% of pediatric cases of CRC are associated with a hereditary CRC syndrome. This report details a non-hereditary case of CRC in a 14-year-old young man.

**Case Presentation:**
Mr. WS is a 14-year-old male with a past medical history of ADHD, ODD, and eczema who presented to the Henry Ford Fairlane emergency department with a two-week history of moderate and intermittent RLQ abdominal pain associated with four episodes of nonbilious vomiting, weight loss, and anorexia. The patient reported the pain was worse when lying on his R side and sudden movement. The patient endorsed less than 3 bowel movements per week over the past several weeks. Physical exam demonstrated voluntary guarding and mild RLQ tenderness, however no peritoneal signs.

The differential diagnosis included celiac disease, volvulus, inflammatory bowel disease, and infection which are all conditions that may cause a painful change in bowel habits. Records from the Henry Ford Fairlane ED showed the following: Labs showed an elevated CEA [5.7 ng/mL] and CRP [10.1 mg/L]. Abdominal US showed multiple, mildly enlarged LNs in the RLQ, however the appendix was not visualized. CT abdomen showed dilated bowel in the RUQ and proximal air fluid levels in the R small bowel consistent with a developing small bowel obstruction, as seen in Figure 1.
Pediatric Gastroenterology was consulted and recommended barium enema. Results deter-mined there was an apple core annular constricting mass in the ascending colon just proximal to the hepatic flexure that measured 6.6 x 6 cm, as seen in Figure 2. The mass had markedly narrowed the lumen of the ascending colon which appeared “string like”. Colonoscopy was planned for several days later.

The patient received a colonoscopy with biopsy on the 4\textsuperscript{th} day of admission, which was consistent with lymphoid nodular hyperplasia and a large colonic mass proximal to the hepatic flexure. The colonic mass was noted to be pink and purple, highly vascular, lobular, and extending 5cm into the lumen of the colon. The lymphoid nodular hyperplasia was noted as scattered erythematous nodules with the most extensive distribution localized to the rectum. Tissue sample from colonoscopy was not adequate to make a histological diagnosis of colorectal cancer. Histology showed only submucosa, however the suspicion for cancer remained high.

The patient received right hemicolectomy, Medi-port placement, and mesenteric lymph node biopsy on the 6\textsuperscript{th} day of admission. Intra-operative frozen pathology of the R colonic mass showed signet cells, which is consistent with colonic adenocarcinoma. Intra-operative staging classified the tumor as stage IIIc colon carcinoma [T4, N1, M0]. The patient was then scheduled for follow up with hematology oncology.

Mr. WS was readmitted 8 months after his original diagnosis of CRC for worsening nausea, vomiting, and abdominal pain. MRI showed thickening of the colon along the primary anastomosis of his previous surgery and seeding of the adjacent peritoneum which is consistent with recurrent CRC. He is currently completing a final round of salvage chemotherapy [Irinotecan and 5-fluorouracil].

The patient and family were disappointed with the recurrence of cancer; however, they report good tolerability of the side effects of chemotherapy. The patient is concerned that our management cannot cure his cancer.
Discussion:
CRC occurs with a peak incidence in patients greater than 50 years of age, however younger patients are developing cases. Most younger patients develop left sided cancers [e.g., rectal] and are diagnosed at later stages. The latter gives evidence that younger patients are indeed developing the disease at an earlier age.[3] As many as 35% of these cases are associated with hereditary syndromes [e.g. Familial Adenomatous Polyposis].[2] However, other risk factors also contribute to earlier disease of onset and include hypertension, hyperlipidemia, obesity, alcohol consumption, and poor vitamin D intake.[4] WS had no documented case of CRC in a first degree relative, so what other perceivable risk factors where present? WS is an obese AAM with a BMI near the 95th percentile for his age, which both increased his risk for earlier disease onset.

Genetics play an important role as risk factors in the development of CRC. Less than 5% of total cases can be attribute to the two most common inherited etiologies of CRC, which are Lynch syndrome and familial adenomatosis polyposis.[5] However, high penetrance mutations still play an important role in development of non-hereditary cases considering an approximate 10% of patients with CRC contain genetic evidence of an APC, BRCA1, BRCA2, CDKN2A, or TP53 mutation. [6] Genetic mutations may predispose families to CRC. Current guidelines are to screen at intervals of 5 years in patients with a first degree relative that is diagnosed with CRC before the age of 60. This patient did not have a known hx of CRC in the family, however a genetic cause remains likely considering the patient’s age of onset. This case report provides further support for development of a genetic panel that will detect patients at higher risk of developing CRC. This would allow early intervention with aggressive screening protocols and protective lifestyle modifications.

Symptoms that cause detection of CRC can provide clues for a patient’s prognosis. CRC that is diagnosed by symptoms [e.g. small bowel obstruction] has a higher risk of metastasis [RR = 3.37], recurrence [RR = 2.19], and a death [RR = 3.02]. [7] Tumors that are specifically discovered by obstruction carry a poor prognosis, and typically guide the decision to pursue adjunctive chemotherapy.[8] WS was unfortunately discovered due to symptoms related to a developing small bowel obstruction which unfortunately portends a poor prognosis and increased his risk for recurrence.

In summary, pediatric cases of CRC are rare but can still occur. Although most pediatric CRC cases are not inherited, there is still likely a genetic component that either involves microsatellite instability or mutations in tumor suppressor genes. A genetic panel should be developed to detect patients at high risk of pediatric CRC so that more aggressive screening protocols can prevent a delay in diagnosis. Detection of patients at the time of symptoms is associated with higher levels of recurrence and metastasis.

Table 1: Important Clinical Data

<table>
<thead>
<tr>
<th>Epidemiology</th>
<th>14-year-old AAM</th>
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<tr>
<td>Past medical history</td>
<td>ADHD, ODD, eczema</td>
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<td>Chief complaint</td>
<td>Insidious onset, progressively worsening RLQ pain w/ nonbilious vomiting.</td>
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<td>Physical exam</td>
<td>RLQ tenderness, hyperactive bowel sounds, no peritoneal signs.</td>
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<td>CT abdomen</td>
<td>Dilated small bowel loops with air fluid levels, significant with developing small bowel obstruction.</td>
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<tr>
<td>Barium enema</td>
<td>Apple core annular constricting mass proximal to the hepatic flexure of the ascending colon.</td>
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<tr>
<td>Colonoscopy</td>
<td>Pink and purple, highly vascular, lobular, colonic mass extending 5cm into the lumen of the colon.</td>
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<td>Frozen pathology</td>
<td>Signet ring cells.</td>
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Figure 1. AP View of CT Abdomen. AP view of CT abdomen retrieved from Henry Ford Fairlane ED. Findings show air fluid levels in the R hemidiaphragm proximal to the hepatic flexure and mildly dilated air-filled loops of small bowel in the left hemi-diaphragm. These findings were attributed to a developing small bowel obstruction.

Figure 2: Barium Enema. AP view of Barium Enema. Findings show an apple core annular constricting mass in the ascending colon just proximal to the hepatic flexure that measured 6.6 x 6 cm. The colonic lumen inside the apple core lesion appears like a string.

References:


[3]. Abualkhair WH. Trends in Incidence of Early-Onset Colorectal Cancer in the United States Among Those Approaching Screening Age. JAMA network open.3[1].

[4]. Ward EM. Annual Report to the Nation on the Status of Cancer, Featuring Cancer in Men and...


