Case Report: Colo-colonic Intussusception In a Young Patient with Lynch Syndrome Family History

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Introduction

Lynch syndrome, or hereditary non-polyposis colorectal cancer (HNPCC), is the most common cause of inherited colorectal cancer, accounting for 3% of all cases. It is an autosomal dominant condition characterized by germline mutations, most commonly MLH1, MSH2, MSH6, and PMS2, or deletions in the EPCAM gene, which leads to microsatellite instability (MSI). This elevates the lifetime risk of malignancies such as colorectal, endometrial or ovarian cancer. Early-onset colorectal cancer with ascending colonic lesions are characteristics of lynch syndrome. We are presenting an unusual case of descending colo-colonic intussusception in a young gentleman, who has a significant family history of lynch syndrome.

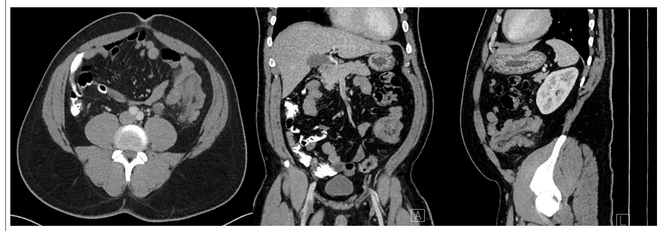
Patient Presentation

A 35-year-old gentleman presented with progressively worsening left lower quadrant abdominal pain for 10 months, as well as intermittent bright red blood per rectum. Admits unintentional weight loss of 20 lbs. Admits intermittent severe constipation but no other complaints. Denies any symptoms of obstruction. Of note, the patient's mother was diagnosed with lynch syndrome at the age of 46, but the patient himself has not been tested.

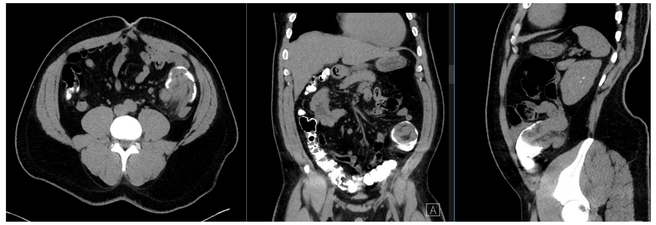
- PMHx: Alcoholic CHF (EF 30-35%), HTN, Hemorrhoids. No surgical hx.
- Vitals: HR 98, RR 19, BP 156/108 (124), T 98.2, Ox Sat 100% on RA
- Exam: Soft, mildly tender to LLQ with palpable mass.
- Labs: WBC 10.1, H/H 13.2/39.2. electrolytes unremarkable.

Imaging

CT scan revealed a 10 cm colocolic intussusception of the descending colon, with 1.7cm circumferential wall thickening of intussuscipiens (Figures 1-3), that is not resolving with 6-hr repeat scan (Figures 4-6). Also found to have a 2.7cm lymphadenopathy. CT chest was negative for suspicious nodules



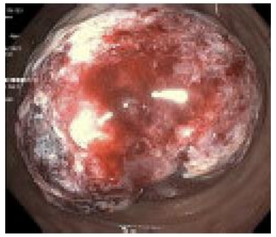
(Figures 1-3) Initial CT A/P with PO and IV contrast in ED: Axial, Coronal and Sagittal view of descending colon intussusception



(Figures 4-6) 6-hour repeat CT A/P: Axial, Coronal and Sagittal view of descending colon intussusception

Hospital course

Flexible sigmoidoscopy was performed on the next day, revealed a partially obstructing tumor at proximal sigmoid colon. The scope could not traverse the mass (Figure 7)
Biopsy of the mass showed invasive adenocarcinoma, G2 with focal mucinous features.



(Figure 7) Partially obstructing mass at the proximal sigmoid

Tumor markers were obtained and turned out to be within normal limits: CEA 2.8.

Patient underwent a laparoscopic procedure converted to open left colectomy with end colostomy creation on hospital day 3.

Surgical pathology revealed Stage IIIB (pT3N1bMx), grade 2 mucinous adenocarcinoma with negative margins; 2 of 15 lymph nodes were positive. Next-generation sequencing identified an MLH1 mutation, consistent with hereditary Lynch syndrome.

The patient recovered uneventfully from surgery and was discharged on postoperative day 4.
The patient is currently undergoing treatment with FOLFOX plus atezolizumab every 2 weeks.

Discussion

Intussusception in Lynch syndrome is exceedingly rare, with fewer than 10 cases reported in PubMed. Most reported cases originate from the ascending colon and result in ileocolic intussusception. Our patient, on the other hand, presented with a descending colocolic intussusception.
Once Lynch syndrome is diagnosed, family members should undergo genetic evaluation and screening based on the Amsterdam II criteria (3-2-1 rule). In retrospect, our patient met criteria for earlier genetic testing given his mother's diagnosis of colorectal cancer at the young age of 46.
Appropriate surveillance for associated malignancies, including colorectal, endometrial, ovarian, gastric, and urinary tract cancers, is essential after the diagnosis of Lynch syndrome.
Lastly, our patient is receiving FOLFOX with atezolizumab based on the ATOMIC trial (Alliance A021502), which evaluated the addition of atezolizumab to adjuvant chemotherapy in patients with MSI-H or dMMR stage III colon cancer. Compared with standard FOLFOX alone, the addition of immunotherapy showed significant improvement of 3-year disease-free survival and reduced recurrence or death by 31%.

References

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