CASE REPORT

Delayed diagnosis of familial adenomatous polyposis in an adolescent patient with a coexisting eating disorder

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SUMMARY

Colorectal carcinoma in the population aged less than 20 years of age is rare but associated with poor prognosis, which is attributable to advanced disease at presentation and higher incidence of the unfavourable mucinous histology. Colorectal carcinoma commonly presents with non-specific gastrointestinal symptoms in conjunction with iron deficiency anaemia. Many of these symptoms can mimic eating disorders, which are common in adolescent women. We present the case of a 20-year-old woman with previously undiagnosed familial adenomatous polyposis and colorectal carcinoma who experienced a significant delay in diagnosis, given a coexisting eating disorder mimicking her symptoms. After confirmation of the diagnosis by colonoscopy and genetic testing, the patient underwent a successful proctocolectomy and experienced full recovery. This case is a reminder that underlying organic pathology should always be excluded prior to a diagnosis of an eating disorder.

BACKGROUND

Although colorectal carcinoma (CRC) is the third most common cancer in adults, the incidence of CRC in the population aged less than 20 years of age is as low as about one per million.¹ Owing to its rarity in this age group, CRC is frequently not considered initially when young patients present with similar symptoms of CRC compared with those in adults.¹ Multiple clinical series have shown that children and young adults with CRC tend to have poorer prognosis compared with adults, which might be attributable to advanced-stage disease at presentation and a higher incidence of aggressive mucinous histology.^{2,3}

CRC commonly presents with non-specific gastrointestinal symptoms in conjunction with iron deficiency anaemia (IDA).^{4,5} Many of these symptoms can mimic eating disorders, which is common in adolescent women. Here we present the case of a 20-year-old woman with previously undiagnosed familial adenomatous polyposis (FAP) and CRC who presented with abdominal discomfort, weight loss and chronic IDA. She experienced a significant delay in diagnosis, given a coexisting eating disorder mimicking her symptoms.

CASE PRESENTATION

A 20-year-old Caucasian woman presented to the emergency department of our hospital reporting symptoms of intermittent nausea and cramping

abdominal pain over the preceding 2 weeks and a single bout of non-bloody emesis on the previous day. The patient's mother also related a distant history of explosive nocturnal diarrhoea. Though the diarrhoea had abated, the patient described a feeling of tenesmus and excessive gas with bowel movements. She denied haematochezia or melena. Four years previously, the patient was diagnosed with IDA for which she inconsistently took iron pills and had experienced profound general weakness and a nearly 40 lbs weight loss over the past 4 years. She admitted a long history of voluntary food restriction but denied purging or bulimic behaviour. Her menses were regular and without menorrhagia or oligomenorrhoea. Other medical history includes tonsillectomy during childhood, gastro-oesophageal reflux disease and vaginal infections. She was not sexually active, nor had she ever had a pelvic examination. She denied the use of alcohol, tobacco or illicit substances. Family history was positive for anorexia in her mother, myocardial infarctions in her father and a distant history of colon cancer in her great-grandfather. Apart from her iron tablets, she occasionally took Excedrin for headaches.

Physical examination of the patient demonstrated a pale and frail woman with no acute distress. She was afebrile, tachycardiac and normotensive. Her body mass index was 16.9 kg/m². Her scalp showed signs of alopecia and her skin fine lanugo hair. Bowel sounds were present and her abdomen was soft, non-distended and non-tender. No abdominal masses or lymphadenopathy were palpated. The laboratory test revealed a significant microcytic anaemia with a haemoglobin of 6.6 g/dL and mean corpuscular volume 61 fL, leukocytosis 18 000/μL with a left shift, platelets 745 000/μL and potassium 3.2 mmol/L and albumin 2.5 g/dL. The iron panel showed significant iron depletion. Folate and B₁₂ were within normal limits. Haemoglobin electrophoresis, antigliadin and endomysial antibodies and transvaginal ultrasound were negative. Gastroenterology consultation was obtained but no endoscopy was performed due to lack of overt bleeding. Given her malnutrition and physical signs of anorexia, nutrition and psychiatry were also consulted. During the hospitalisation, the patient received two units of red blood cell transfusion and electrolyte disturbances were corrected. She was stabilised and discharged with instructions to resume oral iron therapy and to follow up with her primary care physician and haematology for an outpatient workup of her anaemia.

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Following discharge, the patient developed severe nausea and vomiting. She saw her primary care physician 3 weeks later, with an additional 15 lbs weight loss. A digital rectal examination was performed and an irregular mass was palpated. During the examination, the patient collapsed in the office and was returned to the hospital. After being stabilised and aggressively hydrated, the patient underwent colonoscopy the next day, which revealed a myriad of polyps in the rectosigmoid of a variety of sizes, villous appearing changes and possible malignant changes in the sigmoid colon, consistent with familial polyposis. The FAP gene mutation screen confirmed a deletion at the 5' end of exon 15 of the APC gene on chromosome 5. Colorectal surgery was consulted and advocated for total colectomy. The patient was amenable to this, and after a week of outpatient total parenteral nutrition she returned for the procedure. The patient underwent a successful proctocolectomy with ileoanal J-pouch anastomosis and rectal mucosectomy with loop ileostomy. Pathological analysis of the colon confirmed the presence of mucinous adenocarcinoma arising out of greater than 100 adenomatous polyps. Staging determined that the cancer was limited to the submucosa without involvement of the distal margins or sampled lymph nodes.

OUTCOME AND FOLLOW-UP

The patient gained 30 lbs over 1 year after the surgery and her IDA has resolved completely. She has been doing well in follow-up.

DISCUSSION

Although CRC can occur at any age, it is a rare malignancy in the paediatric and adolescent population.^{6 7} The most commonly reported symptoms of CRC in the young age group are persistent abdominal pain, acute intestinal obstruction, bloody rectal discharge, emesis, tenesmus and alteration of bowel habits, anorexia, weight loss, paleness and IDA.^{4 5} Most reviews stated that up to 10% of childhood CRC have predisposing factors such as FAP and its variants (Turcot and Gardners), Peutz-Jegher's, ulcerative colitis, Bloom's syndrome and lynch syndrome, the most common and the best understood condition being Lynch syndrome and FAP.^{6 8}

One cohort study of children and adolescents with CRC showed that the duration of symptoms before presentation ranges from 1 day to 18 months with a median of 3 months and the diagnosis was made 2–6 months after the initial presentation.⁵ The delay of diagnosis helps explain why most of the adolescent patients had advanced disease (Duke class C and D) at presentation and portend a worse prognosis.^{1 5} Besides the delay of diagnosis, the other factor that favours a poorer prognosis of CRC in the younger population is the aggressive and poorly differentiated mucin-producing adenocarcinoma in over 50% patients, which is found in only 11–13% of the adult disease.^{5 9}

To our knowledge, there is no similar reported case in the English literature. Like most adolescents with CRC, our patient experienced a substantial delay in the diagnosis. It is possible that FAP and eating disorder contributed to her IDA which was diagnosed 4 years previously. At presentation, she manifested many of the common symptoms of CRC including abdominal discomfort, weight loss, anorexia and alteration of bowel habits. Complicating her diagnosis was the overlap of many of these symptoms with eating disorders, especially given her age, gender, history of food restriction and physical symptoms of lanugo hair and alopecia. In fact, many gastrointestinal diseases resemble eating disorders in their presentation. It has been

reported that patients with inflammatory bowel disease, peptic acid disease and intestinal motility disorders voluntarily restrict food in a state that resembles anorexia.¹⁰ Stacher *et al*¹¹ describe a case of a young woman with regurgitation and weight loss who was labelled with anorexia nervosa but who was later diagnosed with achalasia. Misinterpretation of gastrointestinal symptoms may be a result of inadequate history taking and bias of age, gender and emaciation. Though physical signs of anorexia were present, our patient's lanugo hair and alopecia are neither sensitive nor specific for eating disorders.^{12 13} More telling for cancer was the presence of long-standing IDA. It is a common clinical manifestation of CRC especially in women and large right-sided tumours.¹⁴ In fact, most patients with a new diagnosis of colorectal cancer are iron deficient at presentation.¹⁵ IDA in combination with gastrointestinal symptoms demands a full evaluation, regardless of matter the patient's age.¹⁶ Therefore, a good history and application of strict diagnostic criteria for eating disorders should always be combined with a high index of suspicion for underlying organic pathology.

Digital rectal examination (DRE) is a relatively simple and inexpensive procedure that is used to examine the lower rectum and other pelvic organs. Although the current guidelines do not include DRE as a routine method for screening colorectal cancer due to its low sensitivity,¹⁷ as shown in this case, DRE was effective in detecting tumours in the rectum, which is within the examiner's reach. In patients with suggestive symptoms, a DRE followed by endoscopy and biopsy is recommended if rectal lesions are suspected.¹⁸ Previous studies identified that gastrointestinal tract as the source of chronic blood loss in a substantial proportion of premenopausal women with IDA; therefore, gastrointestinal endoscopy should be performed in premenopausal female patients with IDA if they present with gastrointestinal symptoms, faecal occult blood, weight loss or menstrual blood loss is not in proportion with the severity of their IDA.^{19–21} In our case, DRE and gastrointestinal endoscopic evaluation were indicated but they were significantly delayed due to the false attribution of the alarming symptoms to the coexisting eating disorder. This case reemphasises the importance of performing a DRE followed by gastrointestinal endoscopy in patients with rectal symptoms, IDA and weight loss.

Learning points

- ▶ Colorectal cancer and eating disorders can present with non-specific gastrointestinal symptoms and iron deficiency anaemia.
- ▶ Underlying organic pathology should always be considered and excluded prior to a diagnosis of an eating disorder.
- ▶ Iron deficiency anaemia in combination with gastrointestinal symptoms demands a full evaluation, regardless of the patient's age.
- ▶ A digital rectal examination followed by gastrointestinal endoscopy should be performed in patients with rectal symptoms, iron deficiency anaemia and weight loss.

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Competing interests None.

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