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CASE REPORT

젊은 환자에서 이중풍선소장내시경으로 진단한 소장암 1예

최윤지, 정성우, 엄준원[1], 이응석[2], 구자설, 임형준, 이상우, 최재현
고려대학교 의과대학 내과학교실, 외과학교실[1], 병리학교실[2]

Small Bowel Carcinoma in Young Patient Detected by Double-balloon Enteroscopy

Yoon Ji Choi, Sung Woo Jung, Jun Won Um[1], Eung Seok Lee[2], Ja Seol Koo, Hyung Joon Yim, Sang Woo Lee and Jai Hyun Choi
Departments of Internal Medicine, Surgery[1] and Pathology[2], Korea University College of Medicine, Seoul, Korea

A 17-year old female presented with a chief complaint of melena and epigastric pain. She had a family history of colon cancer, her mother having been diagnosed with hereditary nonpolyposis colorectal carcinoma (HNPCC). After close examination including double-balloon enteroscopy, the patient was diagnosed with small bowel carcinoma, in spite of her young age. Here we report this rare case of small bowel carcinoma in a young patient with a family history of HNPCC. **(Korean J Gastroenterol 2011;58:217-220)**

Key Words: Jejunal neoplasm; Hereditary nonpolyposis colon cancer; Double-balloon enteroscopy

INTRODUCTION

Small bowel carcinoma is a very rare disease accounting for only 2 percent of all gastrointestinal neoplasms and less than 0.4 percent of all cancers. However, an increased risk of small bowel carcinoma is seen in hereditary nonpolyposis colorectal carcinoma (HNPCC). HNPCC, also known as Lynch syndrome, is a colorectal cancer susceptibility syndrome with an extremely high risk of not only colorectal carcinoma, but also several extracolonic malignancies, including small bowel carcinoma.[1,2]

According to the American Gastroenterological Association (AGA) guidelines, people with familial risk of HNPCC should undergo colonoscopy every 1-2 years beginning at age 20-25 or 10 years younger than the youngest diagnosis of colon cancer in the family.[3] However, there is no recommendation for the surveillance of small bowel tumors in HNPCC patients.

We describe a case of small bowel carcinoma in a young patient with a family history of HNPCC.

CASE REPORT

A 17-year-old female visited the emergency room with complaints of melena and epigastric pain. She had no significant medical history, did not smoke, drink alcohol, or take any medications, but had a family history of colon cancer. Her maternal grandmother had a history of gastric cancer and colon cancer, and her mother was diagnosed with colon cancer and underwent total colectomy at age of 30. The patient's mother had undergone genetic testing for HNPCC, resulting in the detection of a relevant mutation. Although which type of genetic mutation her mother had was not known, the patient was considered to have a strong possibility of HNPCC carrier.

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교신저자: 정성우, 425-707, 안산시 단원구 고잔1동 516, 고려대학교 안산병원 소화기내과
Correspondence to: Sung Woo Jung, Division of Gastroenterology, Department of Internal Medicine, Korea University Ansan Hospital, 516, Gojan 1-dong, Danwon-gu, Ansan 425-707, Korea. Tel: +82-31-412-4854, Fax: +82-31-412-5582, E-mail: sungwoojung@korea.ac.kr

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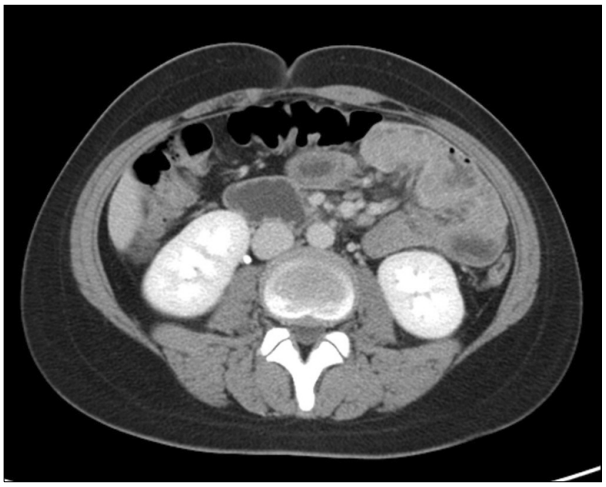

Fig. 1. Abdominal CT finding. It revealed segmental eccentric wall thickening of the jejunum which indicated a pathology of lymphoma or adenocarcinoma.

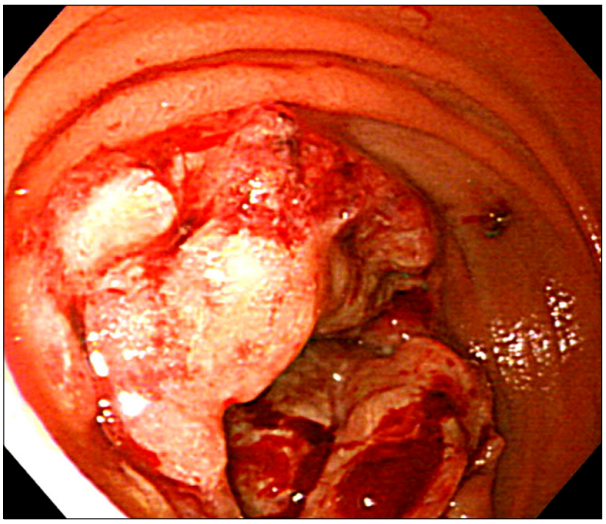

Fig. 2. Endoscopic finding. A large cancerous lesion with friable bleeding mucosa was found in the proximal jejunum that almost obstructed the bowel.

Upon physical examination, the patient's body temperature was 36.7°C, her pulse rate was 96/min, her respiratory rate was 16/min and her blood pressure was 100/60 mmHg. Epigastric tenderness (but no rebounding tenderness) was noted during the abdominal examination. Laboratory tests revealed that the patient had severe iron deficiency anemia. Her white blood cell count was 5,710/mm^3, hemoglobin was 6.2 g/dL and platelets were 484,000/mm^3. Serum iron was 8 μg/dL, total iron binding capacity was 360 μg/dL, ferritin was 6.20 μg/mL (transferrin saturation was 2%), and a stool occult blood test was positive.

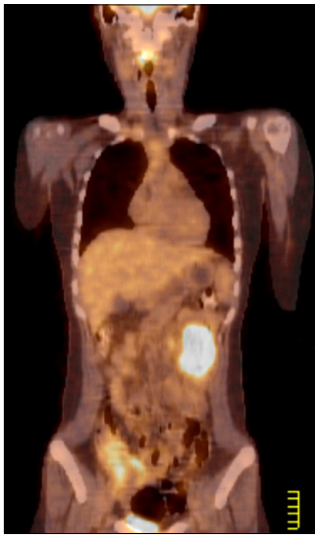

Fig. 3. PET scan finding. It revealed that the jejunal mass showed intense hypermetabolism. There were no other abnormal metabolic lesions.

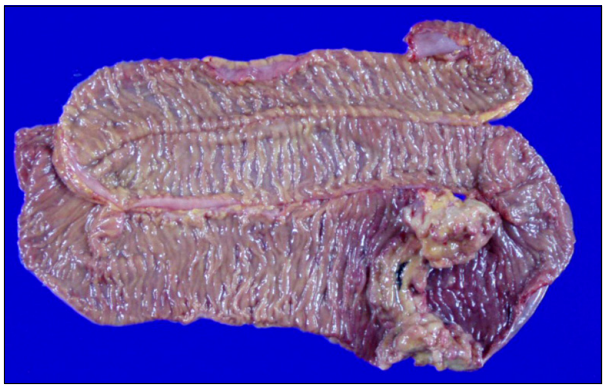

Fig. 4. Gross finding of the jejunal segmentectomy specimen. There was 9.0×5.5 cm mass involving the proximal jejunum.

On suspicion of gastrointestinal bleeding, the patient underwent gastrofiberscopy and colonofiberscopy. There was no bleeding focus. Abdominal CT revealed segmental eccentric wall thickening of the jejunum (Fig. 1). Double-balloon enteroscopy detected a cancerous lesion in the proximal jejunum (Fig. 2) and diagnosis of adenocarcinoma was confirmed pathologically. PET scans revealed a large jejunal mass showing intense fluorodeoxy glucose activity without distant metastasis (Fig. 3).

The patient underwent segmental resection of the jejunum. The resected mass measured 9.0×5.5 cm (Fig. 4) and was pathologically confirmed as adenocarcinoma. The cancer extended to the muscle layer and periintestinal adipose tissue

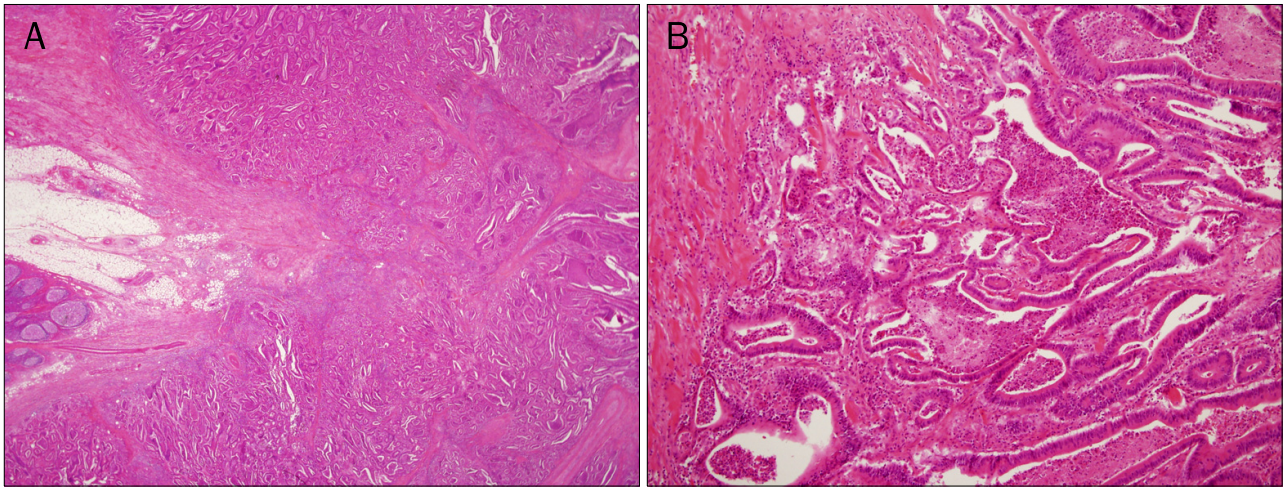

Fig. 5. Pathologic finding. (A) Well differentiated adenocarcinoma extended to the muscle layer and periintestinal adipose tissue of jejunum (H&E, ×12.5). (B) Proliferation of atypical glands with irregular contours. The adenocarcinoma invaded the adjacent tissue with necrosis (H&E, ×100).

of jejunum without invasion into lymph nodes or distant metastasis (Fig. 5). As diagnosed with stage IIa, no additional treatment was needed. Because, adjuvant chemotherapy after surgery for small bowel carcinoma has not been proven beneficial.

The patient was discharged without any complications. Currently, she is healthy and regularly visits outpatient clinics for follow-up and can proceed with abdominal CT scan when needed. And colonoscopy is planned for the patient every 1-2 years to screen for colon cancer.

DISCUSSION

HNPCC is the most common colorectal cancer susceptibility syndrome, with an autosomal dominant mode of inheritance with incomplete penetrance. HNPCC accounts for about 5% of all colorectal cancers and HNPCC patients experience a lifetime risk of colon cancer of about 80%.[1,2] HNPCC is characterized by early onset, predominantly proximal colon carcinomas, excess synchronous and metachronous colorectal carcinomas, and extracolonic malignancies including endometrial, ovarian, gastric, urinary, hepatobiliary, brain, skin, and small bowel.[4] Of these, the most common extracolonic malignancy is endometrial cancer (occuring in 40-60% of female mutation carriers), followed by ovarian cancer (occuring in 12-15% of female mutation carriers).[3]

Small bowel carcinoma is very rare in the general population. However, HNPCC patients are at an increased risk of small bowel carcinoma, with the lifetime risk of small bowel carcinoma in HNPCC patients calculated to range from 1-4%, which is $>$100 times the risk in the general population.[2,4-6]

Due to the high risk of developing cancer, surveillance is recommended for HNPCC patients. Colonoscopic screening has been shown to significantly improve survival among HNPCC patients and mutation carriers.[3] According to AGA guidelines, people with a familial risk of HNPCC should undergo colonoscopy every 1-2 years beginning either at age 20-25 or 10 years younger than the youngest diagnosis of colon cancer in the family.[7] However, no specific recommendations are outlined for other, less common tumors, including tumors of the small bowel.

In our case, colonoscopy was planned for the patient every 1-2 years beginning at age 20 because her mother had been diagnosed with colon cancer at age 30. Nevertheless, the patient was diagnosed small bowel cancer at age 17, before the first surveillance for colorectal cancer. It was the patient's first and only tumor.

Small bowel carcinoma in HNPCC is poorly characterized, and its significance remains unclear. Two large studies have assessed the characteristics of small bowel carcinoma in HNPCC. These studies show that most small bowel tumors found in HNPCC patients are adenocarcinomas with lymphocyte infiltration and expansive growth pattern of the tumor border forming pseudocapsule, and with an anatomic distribution favoring proximal lesions, an earlier average age at onset (compared with 65 years in general population), and better long-term survival rates than small bowel carcinomas in the general population.[8,9]

Small bowel cancer in HNPCC is mostly located in the duodenum or jejunum. Therefore, duodenoscopy or enteroscopy might be beneficial for the early detection of small bowel carcinomas in HNPCC patients. If neither is available, capsule endoscopy may be beneficial.[9,10]

In conclusion, HNPCC patients have very high risk of colorectal carcinoma and other extracolonic tumors. Colonoscopy and other surveillance techniques are strongly recommended after age 20-25. On the other hand, the surveillance of small bowel carcinoma may not be recommended due to its low incidence. However, if an HNPCC affected patient has unexplained anemia or abdominal symptoms that are highly suspicious of small bowel lesions, early enteroscopy or capsule endoscopy as well as a colonoscopy should be considered, because the small bowel carcinoma might be a first manifestation, leading to an early diagnosis.

REFERENCES

1. Aarnio M, Mecklin JP, Aaltonen LA, Nyström-Lahti M, Järvinen HJ. Life-time risk of different cancers in hereditary non-polyposis colorectal cancer (HNPCC) syndrome. Int J Cancer 1995; 64:430-433.
2. Hampel H, Stephens JA, Pukkala E, et al. Cancer risk in hereditary nonpolyposis colorectal cancer syndrome: later age of onset. Gastroenterology 2005;129:415-421.
3. Lindor NM, Petersen GM, Hadley DW, et al. Recommendations for the care of individuals with an inherited predisposition to Lynch syndrome: a systematic review. JAMA 2006;296:1507-1517.
4. Anaya DA, Chang GJ, Rodriguez-Bigas MA. Extracolonic manifestations of hereditary colorectal cancer syndromes. Clin Colon Rectal Surg 2008;21:263-272.
5. Lynch HT, Smyrk TC, Lynch PM, et al. Adenocarcinoma of the small bowel in lynch syndrome II. Cancer 1989;64:2178-2183.
6. Schulmann K, Engel C, Propping P, Schmiegel W. Small bowel cancer risk in Lynch syndrome. Gut 2008;57:1629-1630.
7. Winawer S, Fletcher R, Rex D, et al; Gastrointestinal Consortium Panel. Colorectal cancer screening and surveillance: clinical guidelines and rationale-Update based on new evidence. Gastroenterology 2003;124:544-560.
8. Rodriguez-Bigas MA, Vasen HF, Lynch HT, et al. Characteristics of small bowel carcinoma in hereditary nonpolyposis colorectal carcinoma. International Collaborative Group on HNPCC. Cancer 1998;83:240-244.
9. Schulmann K, Brasch FE, Kunstmann E, et al; German HNPCC Consortium. HNPCC-associated small bowel cancer: clinical and molecular characteristics. Gastroenterology 2005;128: 590-599.
10. Baichi MM, Arifuddin RM, Mantry PS. Metachronous small bowel adenocarcinomas detected by capsule endoscopy in a patient with hereditary nonpolyposis colorectal cancer. Dig Dis Sci 2007;52:1134-1136.