FAMILIAL JUVENILE POLYPOSIS COLI : A CASE REPORT

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Familial juvenile polyposis coli (JPC) is an uncommon autosomal dominant trait characterized by the occurrence of numerous polypoid hamartomas in the colorectum. The condition usually occurs in childhood with high malignant potential. Rectal bleeding, anemia and rectal prolapse are the most common clinical presentations. Two young sisters with presentations of intermittent rectal bleeding, rectal prolapse and severe anemia revealed hamatomatous colonic polyposis. They were treated by transrectal polypectomy and blood transfusion initially, but the symptoms relapsed frequently. They received restorative proctocolectomy finally. The patients recovered uneventfully after operation and remained well during three years of follow up.

Keywords: **familial juvenile polyposis coli, restorative proctocolectomy**

Juvenile polyposis coli (JPC) is an uncommon condition characterized by numerous hamartomatous polyps throughout the colon. It usually presents with rectal bleeding, anemia and rectal prolapse. More and more reports demonstrated that juvenile polyposis is a premalignant condition and the cumulative risk of colorectal cancer is more than 50 percent.[1,2] We report two sisters with juvenile polyposis coli causing persistent rectal bleeding and severe anemia.

CASE REPORT

A 12 years old girl visited our clinic on January of 1993 with complaint of anorectal painful prolapsed mass for 3 days and intermittent anal bleeding for one year. Digital rectal examination revealed multiple rectal polyps with prolapse (fig.1). A CBC revealed severe anemia (Hb: 7.5 gm/dl). She was admitted and received blood transfusion and a transrectal polypectomy was done. More than 10 polyps, measuring 0.5 to 4 cm in size, were removed. Histologic examination revealed juvenile polyps, characterized by mucus filled dilated cystic colonic glands lined by normal columnar mucosa (fig.2). On each follow up visit, however, she continued to show severe anemia with hemoglobin level around 7gm/dl and needed blood transfusion every time. A colonoscopy was finally performed in October 1996

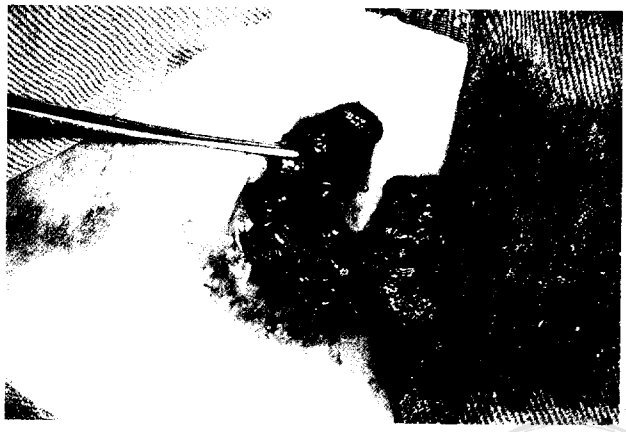

Fig 1. Rectal polyps prolapsed to the anus.

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Fig 2. Microscopically, the juvenile polyps characterized by mucus filled cystically dilated tubules lined by normal inflamed mucosa. (H&E stain, x200)

and numerous polyps, varied in size, were found all over the colon, especially concentrated in the rectum and cecum. Laboratory tests showed severe hypoalbuminemia (albumin level: 1.9 gm/dl). Because of persistent rectal bleeding, intractable anemia and difficulty in eradicating all polyps by endoscopy, a restorative proctocolectomy with ileal J-pouch anal anastomosis and protective loop ileostomy were performed in October 1996. The post-operative course was rather smooth and the hemoglobin and albumin level gradually returned to normal level.

Another 14-year-old girl, the elder sister of the first case, who visited our clinic in July 1994 with the chief complaint of intermittent anal prolapse and bleeding for one year. Laboratory investigations showed severe iron deficiency anemia (Hb: 4.3 gm/dl, Ferritin: 1.93 ng/ml) but her albumin level was normal (3.7 gm/dl). Colonoscopy revealed numerous graph-like polyps in the entire colon, especially over the ascending and descending colon. She was first admitted for blood transfusion and transrectal polypectomy to relieve the symptom of rectal prolapse. About 40 polyps were removed from the rectum. Histopathologic examination of the polyps revealed juvenile polyps. She was discharged uneventfully and had regular follow-up at our clinic. Due to persistent rectal bleeding, anemia requiring blood transfusion, and occasional abdominal cramping pain, suggestive of colonic intussusception, definitive surgical treatment was advised. She underwent

Fig 3. Gross specimen showed multiple polyps over entire colon, especially at cecum and rectum.

restorative protocolectomy with ileal J-pouch anal anastomosis and protective loop ileostomy in November 1995 (fig.3). She was discharged in good condition 14 days after the operation. Her hemoglobin level eventually returned to normal range.

Both cases underwent take-down of ileostomy 6 months after the major operation. The post-operative period were without incidents and the frequency of bowel movement remain acceptable. After 3 and 4 years of follow-up with colonoscopy in each case, no other polyp was found in the gastrointestinal tract. Their little brother had colonoscopy surveillance and was found to have asymptomatic and less extensive colonic polyposis.

DISCUSSION

Juvenile polyp, or retention polyp, is defined as polypoid harmatomas arising from the lamina pro-

pria of the gastrointestinal tract within the pediatric age group.[3] The diagnostic criteria of juvenile polyposis are: (1) three or more colonic juvenile polyposis; (2) juvenile polyps throughout the gastrointestinal tract; (3) any number of juvenile polyps in a patient with a family history of juvenile polyposis.[4] There are three forms of this rare condition in which polyps are limited to the colon (juvenile polyposis coli), distributed throughout the entire gastrointestinal tract (generalized juvenile polyposis), or present in infancy either as colonic or diffuse disease (infantile polyposis syndrome).[5]

In 20 to 50 percent of the cases, JPC occurs as a familial condition with an autosomal dominant trait.[6] Recently, some authors found that a subset of juvenile polyposis families carry germ line mutations in the gene SMAD4(DPC4), located on chromosome 18q21.1.[7,8] Since 1980s, it has been recognized that the polyps of juvenile polyposis have malignant potential.[9-12] The juvenile polyposis have the malignant potential of 9-20 percent while the cumulative lifetime risk of colorectal cancer for familial juvenile polyposis is 50 percent.[1,2]

The most common clinical presentations of JPC include rectal bleeding, anemia and prolapse of either the polyp or the rectum itself as seen in our cases. Other clinical features such as stunted growth, intussusception, diarrhea, protein loss enteropathy, and associated congenital defects had been reported.[13] In our cases, hypoalbuminemia was only found in the younger sister. The majority of patients will present symptoms during the first or second decade but 15 percent of patients with JPC do not have symptom until they are adult.[6] Male preponderance was noted by some authors[4] but this remains controversy.

Macroscopically, the juvenile polyps are 5-50 mm in size, red to brown in color, pedunculated and spherical or lobulated in shape, often show superficial ulceration. Microscopically, the characteristic feature is dilated cystic glands lined by tall columnar epithelium. The lamina propria is expanded and infiltrated by inflammatory cells consisting of neutrophils, eosinophils and a few lymphocytes.[6]

The management of JPC depends on their number and extent, and on the patient's symptoms. Serial colonoscopic polypectomy should be considered first for patients whose symptoms are mild and the polyps can be cleared endoscopically. When polyps are too numerous and difficult to be removed by endoscopy, or when symptoms such as bleeding and diarrhea are intractable, surgery should be considered.[6] Some authors suggests prophylactic colectomy for patients with familial juvenile polyposis coli, since their lifetime risk of cancer development may reach 50 percent.[11] The surgical choices are total colectomy with ileorectal anastomosis or proctocolectomy with pouch. We performed restorative proctocolectomy with J-pouch and ileoanal anastomosis for the sisters to reduce the frequency of diarrhea after the operation. Because some authors reported recurrence of polyps in the ileal pouch after total proctocolectomy for juvenile polyposis,[14] endoscopic surveillance of the gastrointestinal tract should be done periodically. Fortunately, no recurrence was noted in our cases after three years of follow-up. For the first-degree relatives, screening colonoscopy should be done as with familial adenomatous polyposis and should start in the second decade.[4]

In conclusion, although juvenile polyps were regarded as a benign lesions, familial juvenile polyposis coli has high malignant potential. Restorative proctocolectomy is recommended for patients with intractable symptoms or when endoscopic clearance of polyps is impossible. These patients also need continuing endoscopic follow-up to detect any recurrence.

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家族性幼年型大腸息肉症：病例報告

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家族性幼年型大腸息肉症是一種少見的常染色體顯性遺傳疾病，特徵是在大腸直腸有無數息肉性的缺陷瘤，通常發生在小孩，並且有變惡性的可能。常見臨床症狀有肛門出血、貧血及肛門脫出。我們要報告兩姐妹有間歇性的肛門出血及脫出症狀，檢查發現有很多幼年型大腸息肉及嚴重貧血，她們接受經肛門息肉切除術及多次的輸血治療，但症狀仍然持續復發，最後她們分別都接受大腸直腸全切除手術，兩姐妹術後皆恢復良好，且經過三年多的追蹤，並無發現復發情形。

關鍵詞：家族性幼年型大腸息肉症，大腸直腸全切除手術，遺落