Is Colonoscopy Necessary in Children Suspected of Having Colonic Polyps?

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Background/Aims: The clinical spectrum, histology, and endoscopic features of colonic polyps in the pediatric age group were studied to evaluate the role of colonoscopy in children suspected of having colonic polyps. **Methods:** Seventy-six patients with colorectal polyps were studied. Investigations included barium enema (n=6), sigmoidoscopy (n=17), and colonoscopy (n=53) at the initial visit. Colonoscopy was also performed in 23 patients who received barium enema or sigmoidoscopy. Data related to age, gender, family history, signs, symptoms, size, location, polyp types, and associated diseases were collected and analyzed.

Results: Among the 76 patients, juvenile polyps were detected in 58 (76.3%), potentially premalignant polyposis in 17 (22.4%), familial adenomatous polyposis in 11 (14.5%), Peutz-Jegher syndrome in 4 (5.3%), and juvenile polyposis syndrome in 2 (2.6%). Twenty-two patients (28.9%) had polyps in the upper colon. All patients with potentially malignant polyps had polyps in both the upper colon and rectosigmoid colon.

Conclusions: Although most of the children with colorectal polyps had juvenile polyps, a significant number of cases showed multiple premalignant and proximally located polyps. This finding emphasizes the need for a colonoscopy in such patients. Thus, the risk of malignant change, particularly in children with multiple polyps, makes surveillance colonoscopy necessary. (*Gut Liver* 2010;4:326-331)

INTRODUCTION

In children, colorectal polyps are usually accompanied by painless rectal bleeding. Colorectal polyps are common during childhood and affect 1.1% of preschool and school-aged children.¹ More than 90% of such cases are juvenile polyps, and the majority of these occur in the rectosigmoid region. Juvenile polyps are generally solitary and have been considered to be at a little or no malignant risk. However, there are cases on record of adenomatous change or a colorectal adenocarcinoma developing from juvenile polyps in children.²⁻⁶ With the wide use of pediatric colonoscopy, there is an increasing recognition of polyposis syndromes in children, including the hereditary polyposis syndrome, with its well known malignant potential.

Most children undertake a sigmoidoscopy or barium enema when they are suspected to have a colonic polyp, because colonoscopy appears to be very invasive in children. However, there is a lower chance of finding out whether the patient has polyps with malignant potential if only sigmoidoscopy or barium enema without double contrast is performed in younger children. In the present study, the role of colonoscopy in children with suspected colonic polyps was evaluated.

MATERIALS AND METHODS

1. Materials

The medical records of 76 children diagnosed with colorectal polyps from January 1998 to April 2009 at the Samsung Medical Center were retrospectively reviewed.

Key Words: Children; Colonoscopy; Colonic polyps

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Data related to age, gender, family history, signs and symptoms, size, location, type of polyp, and associated diseases were collected and analyzed. The study was approved by the institutional review board of Samsung Medical Center.

2. Methods

Seventy-six patients with colorectal polyps were studied. The investigations included barium enema (n=6), sigmoidoscopy (n=17), and colonoscopy (n=53) at the initial visit. Colonoscopy was additionally performed in 23 patients who previously underwent barium enema or sigmoidoscopy.

Detailed family histories of colorectal polyps or cancer were obtained from all patients studied. Familial adenomatous polyposis (FAP), Peutz-Jegher syndrome, and juvenile polyposis syndrome were categorized as premalignant polyposis because they are well known to have malignant potential.

FAP was diagnosed by the presence of multiple adenomatous polyps in families with known FAP,⁷ and genetic testing was performed in these patients. Turcot syndrome was included in FAP. The diagnosis of the Peutz-Jegher syndrome was made based on the presence of multiple gastrointestinal hamartomatous polyps and the melanin pigmentation of the skin and the mucous membranes.⁷ The diagnosis of the juvenile polyposis syn-

drome was made when one of the following criteria was met: 1) more than five juvenile polyps of the colon or rectum, 2) juvenile polyps in other parts of the gastrointestinal tract other than the colon, or 3) any number of juvenile polyps and a positive family history.^{4,7}

RESULTS

1. Classification of colorectal polyps

Table 1 shows the clinical characteristics of the patients with colorectal polyps. There were juvenile polyps in 58 (76.3%) and potentially malignant familial adenomatous polyposis, Peutz-Jegher syndrome, and juvenile polyposis in 17 (22.4%). The clinical characteristics of the patients with premalignant polyposis are shown in Table 2. Juvenile polyposis was diagnosed in 2 (2.6%) and Peutz-Jegher syndrome in 4 (5.3%). In 11 (14.5%) cases, diagnosis of familial adenomatous polyposis was made on those with more than 100 multiple adenomatous polyps. Two of these 11 patients had been previously confirmed to have a medulloblastoma and were diagnosed with the Turcot syndrome. One patient was diagnosed with a villous adenoma, 17 patients with potentially malignant polyps, 5 (28.9%) and 22 of the 58 respectively had juvenile polyps and polyps in the upper colon. All patients with potentially malignant polyps had polyps in the upper colon as well as in the rectosigmoid colon.

2. Juvenile polyps

1) Age and gender distribution of juvenile polyps

The mean age of the patients with juvenile polyps was 6.6 years with a male-to-female ratio of 1.9:1 in 38 male

Table 1. Clinical Characteristics of Patients with Colorectal Polyps

Sex, n	
Male	46
Female	30
Age	
Mean	8.5 yr
Range	9 mo to 24 yr
Diagnosis, n (%)	
Juvenile polyp	58 (76.3)
FAP	11 (14.5)
Peutz-Jegher syndrome	4 (5.3)
Juvenile polyposis syndrome	2 (2.6)
Villous adenoma	1 (1.3)
Investigation at initial visit, n (%)	
Barium enema	6 (7.9)
Sigmoidoscopy	17 (22.4)
Colonoscopy	53 (69.7)
Initial symptoms and signs, n (%)	
Hematochezia	66 (86.8)
Diarrhea	8 (10.5)
Prolapse of polyp	4 (5.3)
Abdominal pain	2 (2.6)
Constipation	1 (1.3)

FAP, familial adenomatous polyposis.

Table 2. Clinical Characteristics of Patients with Premalignant Polyposis, Familial Adenomatous Polyposis, Peutz-Jegher Syndrome, and Juvenile Polyposis Syndrome

Sex, n	
Male	8
Female	9
Age	
Mean	15.0 yr
Range	9 mo to 24 yr
Investigation at initial visit, n (%)	
Barium enema	1 (5.9)
Sigmoidoscopy	3 (17.6)
Colonoscopy	13 (76.5)
Initial symptoms and signs, n (%)	
Hematochezia	8 (47.0)
Diarrhea	7 (41.2)
Lip pigmentation	3 (17.6)
Family history of polyps	2 (11.8)
Prolapse of polyp	1 (5.9)

and 20 female children, indicating a predominance of males with juvenile polyps (Fig. 1).

2) Clinical characteristics of patients with juvenile polyps

Among the 58 patients with juvenile polyps, rectal bleeding was present in 56 (96.5%). In two children without rectal bleeding, prolapse of the polyp was noted in one, and the other presented abdominal pain. The symptom duration of rectal bleeding varied from several days to one year.

3) Morphological and histological characteristics of juvenile polyps

Among the 58 patients with juvenile polyps, a single polyp was found in 56 (96.5%) and 2 polyps each in 2 (3.5%). The sizes of the polyps were: 5 mm or smaller in 9 (15%), 6-10 mm in 16 (26.7%), 11-20 mm in 28 (46.7%), 21-20 mm in 6 (10%), as well as 1 case (1.7%) with a large polyp of 50 mm (Table 3).

Juvenile polyps were most frequently found in the rectum in 40 (66.7%) patients, and 15 (25%) in the sigmoid

Fig. 1. Age distribution of patients with juvenile polyps.

Table 3. Number and Size of Juvenile Polyps

	No. (%)
No. of polyp per patient (n=58)	
1	56 (96.5)
2	2 (3.5)
≥3	0 (0)
Size, mm (n=60)	
≤5	9 (15.0)
6-10	15 (26.7)
11-20	26 (46.7)
21-30	6 (10.0)
>30	1 (1.7)

colon; most of the juvenile polyps occurred in the rectosigmoid colon. One (1.7%) each polyp was found in the descending, transverse, and ascending colons, and 2 (3.3%) were detected in the cecum. With regard to the shape: 42 cases were pedunculated polyps (70%) and 18 were sessile polyps (30%) (Table 4).

3. Juvenile polyposis

Two patients were diagnosed with juvenile polyposis. One was a 16-year-old female with no rectal bleeding; her father previously had a colonoscopy and was diagnosed with rectal cancer. Multiple polyps (23) were found by colonoscopy in this patient. In addition, juvenile polyps were found by polypectomy, and adenomatous changes were also noted.

The other patient was a 7-month-old female with rectal bleeding and polyps separated from the intestinal wall. This patient had no family history of polyps. Initially, when she underwent sigmoidoscopy, 2 polyps were detected. After additional colonoscopy, approximately 70-80 polyps, 3-10 mm, were noted in all regions of the colon. The diagnosis of juvenile polyps was made by biopsy. Both SMAD4 and BMPR1A mutation analyses of this patient gave negative results.

4. Peutz-Jegher syndrome

Four cases of Peutz-Jegher syndrome were found from a 4-, 6-, 8-, and 12-year-olds; all four patients had family histories of the Peutz-Jegher syndrome. Intussusception occurred in two of the patients during the follow-up. Segmental resection of the jejunum and end-to-end anastomosis were performed on one patient, and surgical reduction and lesion removal at the lead point on the other.

5. Familial adenomatous polyposis

A total of 11 children, 4 males and 7 females, were diagnosed with familial adenomatous polyposis, at a mean age of 19 years. A family history of familial adenomatous

Table 4. Location and Type of Juvenile Polyps

	No. (%)
Location of polyp (n=60)	
Rectum	40 (66.7)
Sigmoid	15 (25.0)
Descending colon	1 (1.7)
Transverse colon	1 (1.7)
Ascending colon	1 (1.7)
Cecum	2 (3.3)
Type of polyps (n=60)	
Pedunculated	42 (70)
Sessile	18 (30)

Table 5. Characteristics of Patients with Familial Adenomatous Polyposis

Sex, n	
Male	4
Female	7
Age, yr	
Mean	19
Range	12 to 24
Family history, n (%)	
FAP	6 (54.5)
Colorectal cancer	6 (54.5)
None	3 (27.3)

FAP, familial adenomatous polyposis.

polyposis was found in 6 children (54.5%), and a history of colon or rectal cancer in 6 children (54.5%). No family history of polyps or colon cancer was found in 3 children (27.3%) (Table 5).

In 7 cases (63.6%) diarrhea was the most common presenting symptom prompting colonoscopy; other symptoms such as rectal bleeding, abdominal pain and discomfort were also reported (Table 6). In these patients, extra-intestinal lesions were identified in 3 cases, one of whom was diagnosed with a hepatoblastoma at 34 months of age. The other two were diagnosed with the Turcot syndrome presented by a medulloblastoma.

1) Turcot syndrome

One of the two patients, who were diagnosed and treated for medulloblastoma at the age of 16, was diagnosed with Turcot syndrome after the detection of familial adenomatous polyposis by colonoscopy performed due to the persistent diarrhea continuing for over 2 months at 24 years of age. The mother of this patient died of rectal cancer, and the APC mutation was identified in this patient. Screening tests were carried out in other family members, and a 23-year-old sister of this patient was found to have familial adenomatous polyposis. The other patient, who had previously been reported in a Korean journal in 2007⁸ was found not to have the APC mutation.

6. Villous adenoma

The patient with a villous adenoma was a 17-year-old female; a colonoscopy was performed due to rectal bleeding for 5 years. A 2-cm polyp was found 20 cm above the anus, and polypectomy was performed. The diagnosis of villous adenoma was made based on the biopsy results.

Table 6. Reasons for Colonoscopy in Patients with Familial Adenomatous Polyposis

Symptoms & signs	No. (%)
Diarrhea	7 (63.6)
Hematochezia	6 (54.5)
Abdominal pain/discomfort	2 (18.2)
Familial history of FAP	1 (9.1)

FAP, familial adenomatous polyposis.

DISCUSSION

Colonoscopy performed in all 76 patients revealed that not only were the polyps in 28.9% of the subjects located in the upper colon, but also 22.4% of the patients had potentially malignant polyps. These findings emphasize the role of colonoscopy in children who are suspected to have polyps, even though the pediatric colonoscopy is more invasive, compared to the adult cases.

In general, studies reported so far mentioned the predominance of juvenile polyps in males compared to females;^{3,6,9,10} juvenile polyps have been reported to be twice as common in boys compared to girls. However, in one report, juvenile polyps were equally common among both males and females,¹¹ whereas another group¹² reported that they were more common in females. In the present study, juvenile polyps were more common in males with a male-to-female ratio of 1.9:1. The reported mean age of the children with juvenile polyps were between 5.0 and 7.4 years of age,^{3,6,9-13} the mean age of the present study was 6.6 years. In children, juvenile polyps are generally presented as painless, intermittent rectal bleeding with or without recurrent abdominal pain or prolapsed through the anus. Rectal bleeding was the presenting symptom in 96.5% of the cases, and prolapse of a polyp was observed in one case. Other potential symptoms include mucous in the stool and changes in bowel habits such as diarrhea or constipation; however, these symptoms were not detected in the present study.

Until a total colonoscopy was performed on patients suspected of having polyps, it was generally accepted that juvenile colorectal polyps are solitary and that up to 90% are found in the rectosigmoid colon.¹⁴⁻¹⁶ However, in recent studies, two or more polyps were present in 24-58% of patients, with 15-60% of polyps located proximal to the splenic flexure.^{3,6,11,17} A prior study¹⁸ showed that 94.4% of juvenile polyps were found in the rectum and rectosigmoid colon, and 12.5% of the cases had multiple polyps. However, the colonoscopy was performed only on 28% of all cases, and the author was not able to exclude the possibility of juvenile polyposis in 72% of the

patients. In the present study, all patients undertook colonoscopy, which revealed that 3.3% of the patients had multiple juvenile polyps and 8.3% had polyps located in the upper colon. Of the 23 patients who undertook additional colonoscopy after sigmoidoscopy or barium enema, a 7-month-old girl with juvenile polyposis was initially found to have only a couple of polyps by sigmoidoscopy.

Approximately 50% of the individuals with juvenile polyposis develop gastrointestinal cancer.¹⁹ Accordingly, differentiation of the juvenile polyp from the juvenile polyposis syndrome is essential. Genetic counseling is indicated if a child is suspected of having juvenile polyps. SMAD4 or BMPR1A mutation is present in 40% to 60% of patients with juvenile polyposis.²⁰ In this particular study, SMAD4 and BMPR1A testings were performed in only one patient with the juvenile polyposis syndrome, and the mutation was not observed. Approximately 50% of individuals with the juvenile polyposis syndrome develop adenomatous changes. In the present study, adenomatous change was found in one out of two patients. Because the potential risk for malignant change increases from the age of 20, continuous regular evaluation of the 7-month-old patient is required.

Peutz-Jegher syndrome is a rare genetic disorder with an autosomal-dominant mode of inheritance.²¹⁻²³ Patients with the Peutz-Jegher syndrome can present intermittent abdominal pain, intussusception, rectal bleeding, and bowel obstruction. In the present study, abnormal pigmentation was found in 4 patients, and 2 of the patients underwent surgical treatment for intussusception. Forty-eight percent of the affected individuals developed cancer in the intestinal tract or other organ systems.²⁴ Breast, pancreas, and ovarian cancers are common, and testicular and uterine cancer can also occur. Thus, regular follow-up of these patients is also required. Recommendations for cancer surveillance vary, but aggressive screening recommendations suggest performance of upper and lower endoscopies every 2 years beginning at about 10 years of age.²⁵

Familial adenomatous polyposis is the most common polyposis syndrome of children; it is inherited in an autosomal dominant fashion. The prevalence is between 1 out of 17,000 to 1 of 5,000; in the present study, 11 (14.5%) out of the 76 patients with the diagnosis of colon polyps were confirmed to have familial adenomatous polyposis. Familial adenomatous polyposis is caused by a mutation in the adenomatous polyposis coli (APC) tumor suppressor gene on chromosome 5q. Approximately 30% of the cases result from spontaneous mutations of the APC gene.²⁶ In the present study, 3 out of 11 patients confirmed to have familial adenomatous polyposis had no

family history of colon cancer or familial adenomatous polyposis, and 2 had positive results on the APC testing. Although the mean age for the development of colon cancer in patients with FAP is 39 years, there are reports of cases with colon cancer in children as young as 5 years old.²⁷ The lifetime risk for colorectal cancer is 100%, and adenomas can develop in other regions of the gastrointestinal tract (e.g., stomach and duodenum). In addition, these patients were also at a 1% risk of developing a hepatoblastoma during infancy and early childhood. In the present study one patient was identified with a hepatoblastoma.

In patients with familial adenomatous polyposis, various extraintestinal manifestations can be present including desmoid tumors, rhabdomyosarcoma, dental abnormalities, osteoma, brain tumors, thyroid carcinoma, and congenital hypertrophy of the retinal pigment epithelium.²⁸ Turcot syndrome, first described in 1959 by Turcot *et al.*,²⁹ is characterized by the concurrence of polyposis of the colon and the brain tumor. The two major brain tumors associated with the Turcot syndrome are medulloblastoma and glioblastoma.³⁰ Although patients with familial adenomatous polyposis have a higher risk of brain tumor than the general population, the Turcot syndrome is a rare disorder with a frequency of one occurrence per 3,735 person-years.³⁰ In the present study, 2 out of 11 patients with familial adenomatous polyposis had the Turcot syndrome with a medulloblastoma. Medulloblastoma was first detected in one of our cases, and familial adenomatous polyposis was first detected in another patient. There are no guidelines available for the detection of brain tumors in patients with familial adenomatous polyposis. Therefore, it is yet unclear whether all patients with familial adenomatous polyposis should be screened for the Turcot syndrome, which is an extremely rare hereditary disease.

In conclusion, the present study showed that more than one fifth of the colorectal polyps in children have malignant potential. In addition, considering that 28.9% of the subjects and all patients with potentially malignant polyps had polyps in the upper colon as well as in the rectosigmoid colon, the need for colonoscopy is essential in children with suggestive symptoms. The risk for malignant change, particularly in children with multiple polyps, makes surveillance colonoscopy a requirement. However, the findings of the present study are limited to a single tertiary care hospital in Korea. Thus, further study including multiple medical institutions is required.

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