Clinical Case Report

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# Juvenile polyposis syndrome

## An unusual case report of anemia and gastrointestinal bleeding in young infant

Yi-Han Hsiao, MD[a], Chin-Hung Wei, MD[b,c], Szu-Wen Chang, MD[a], Lung Chang, MD[a], Yu-Wei Fu, MD[c], Hung-Chang Lee, MD[a], Hsuan-Liang Liu, PhD[d], Chun-Yan Yeung, MD, PhD[a,b,d,*]

**Abstract**

**Background:** Juvenile polyposis syndrome, a rare disorder in children, is characterized with multiple hamartomatous polyps in alimentary tract. A variety of manifestations include bleeding, intussusception, or polyp prolapse. In this study, we present an 8-month-old male infant of juvenile polyposis syndrome initially presenting with chronic anemia. To the best of our knowledge, this is the youngest case reported in the literature.

**Methods:** We report a rare case of an 8-month-old male infant who presented with chronic anemia and gastrointestinal bleeding initially. Panendoscopy and abdominal computed tomography showed multiple polyposis throughout the entire alimentary tract leading to intussusception. Technetium-99m-labeled red blood cell (RBC) bleeding scan revealed the possibility of gastrointestinal tract bleeding in the jejunum. Histopathological examination on biopsy samples showed Peutz-Jeghers syndrome was excluded, whereas the diagnosis of juvenile polyposis syndrome was established.

**Results:** Enteroscopic polypectomy is the mainstay of the treatment. However, polyps recurred and occupied the majority of the gastrointestinal tract in 6 months. Supportive management was given. The patient expired for severe sepsis at the age of 18 months.

**Conclusion:** Juvenile polyposis syndrome is an inherited disease, so it is not possible to prevent it. Concerning of its poor outcome and high mortality rate, it is important that we should increase awareness and education of the parents at its earliest stages.

**Abbreviation:** HIV = human immunodeficiency virus.

**Keywords:** anemia, gastrointestinal bleeding, infant, juvenile polyposis syndrome

### 1. Introduction

Juvenile polyposis syndrome is a rare intestinal abnormality in children, particularly in infants.[1] It is characterized with hundreds of polyps in alimentary tract. It has been shown that affected children are susceptible to cancers and fatal medical conditions.[2] The common presentations include anemia, recurrent gastrointestinal bleeding, diarrhea, rectal prolapse, intussusception, protein-losing enteropathy, starvation, and malnutrition. We report a case of an infant with juvenile polyposis syndrome.

This study was approved by Mackay Memorial Hospital Institutional Review Board (16MMHIS042E).

### 2. Case report

An 8-month-old male infant presented melena and iron deficiency anemia with hematocrit of 10.6% and hemoglobin of 2.9 g/dL. Blood transfusion was conducted; however, anemia still persisted. Chronic gastrointestinal hemorrhage was impressed. Technetium-99m-labeled red blood cell bleeding scan showed a negative result. Panendoscopy revealed several polypoid lesions with ulceration over the body and the prepyloric area, which were bleeding to touch (Fig. 1). Pathology of the lesion illustrated inflammation and hyperplasia. Computed tomography depicted numerous nodules throughout the entire alimentary tract, indicating intestinal polyposis (Fig. 2). Asymptomatic small intestinal intussusceptions were also noted.

Owing to uncorrectable anemia as well as sonography constantly demonstrating intussusceptions (Figs. 3 and 4), laparoscopy-assisted enteroscopic polypectomy and reduction of intussusceptions were performed (Fig. 5). The polyps in stomach and duodenum were removed by gastroscopy. The patient recovered uneventfully from this episode. The pathology report described hamartomatous polyps with elongation, tortuosity, and dilatation of the gastric foveae and intestinal mucosal glands.[3] The underlying stroma is characterized by broadband smooth muscle fibers, intermingling with the glands (Fig. 6). Genetic analysis was negative for Peutz-Jeghers syndrome.

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[a] Division of Gastroenterology and Nutrition, Department of Pediatrics, Mackay Memorial Hospital, [b] Department of Medicine, Mackay Medical College, [c] Division of Pediatric Surgery, Department of Surgery, Mackay Memorial Hospital, [d] Institute of Biotechnology and Department of Chemical Engineering, National Taipei University of Technology, Taipei, Taiwan.

[*] Correspondence: Chun-Yan Yeung, Division of Gastroenterology and Nutrition, Department of Pediatrics, Mackay Memorial Hospital, Mackay Medical College, No. 92, Sec. 2, Zhongshan N. Rd., Taipei City 10449, Taiwan (e-mail: cyyeung@mmh.org.tw).



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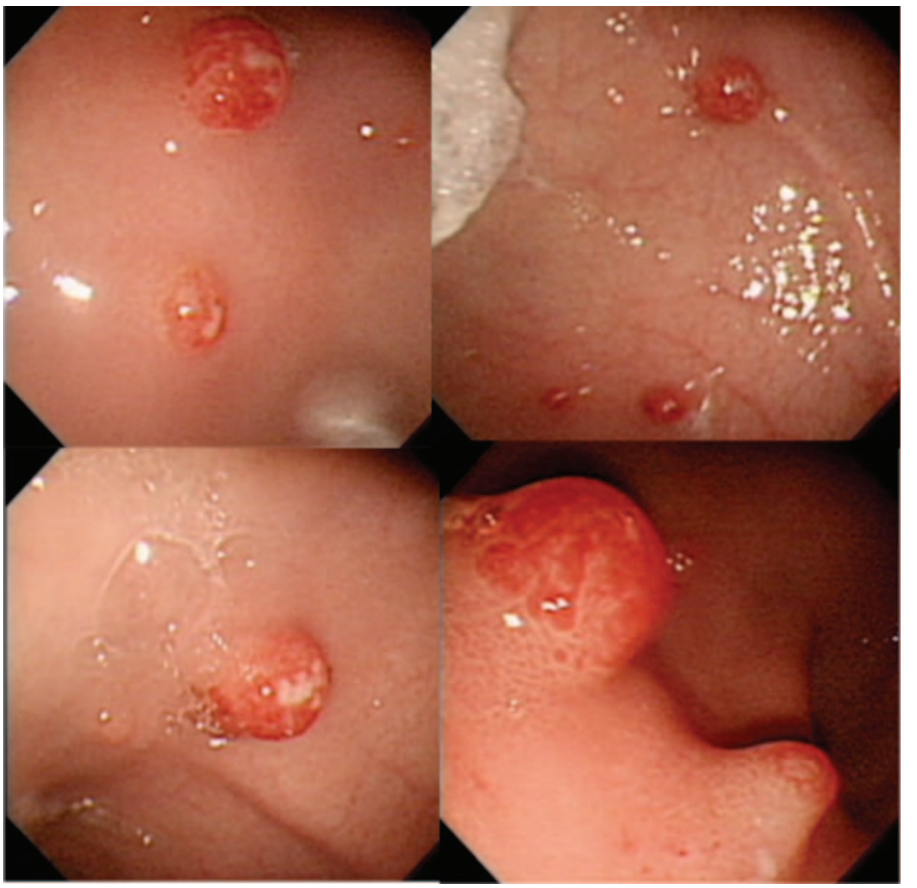

**Figure 1.** Panendoscopy: at least 6 polypoid lesions over gastric body and prepyloric area with ulceration and touch bleeding.

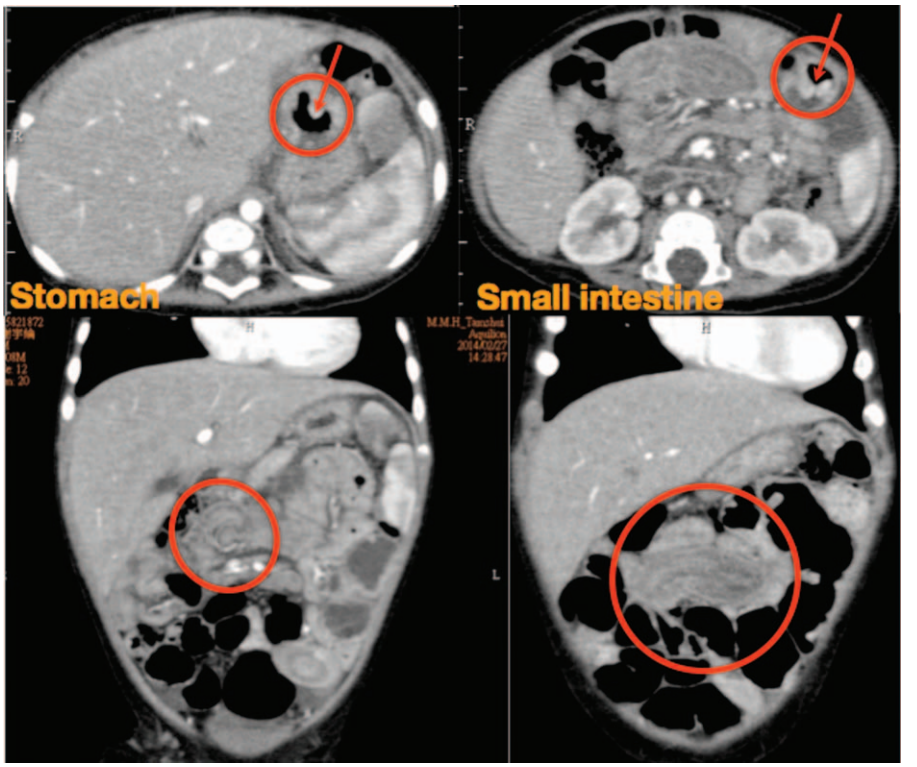


**Figure 2.** Abdominal computed tomography showed multiple nodules within the stomach, small intestine (including duodenum, jejunum, and ileum), and descending colon leading to intussusception.

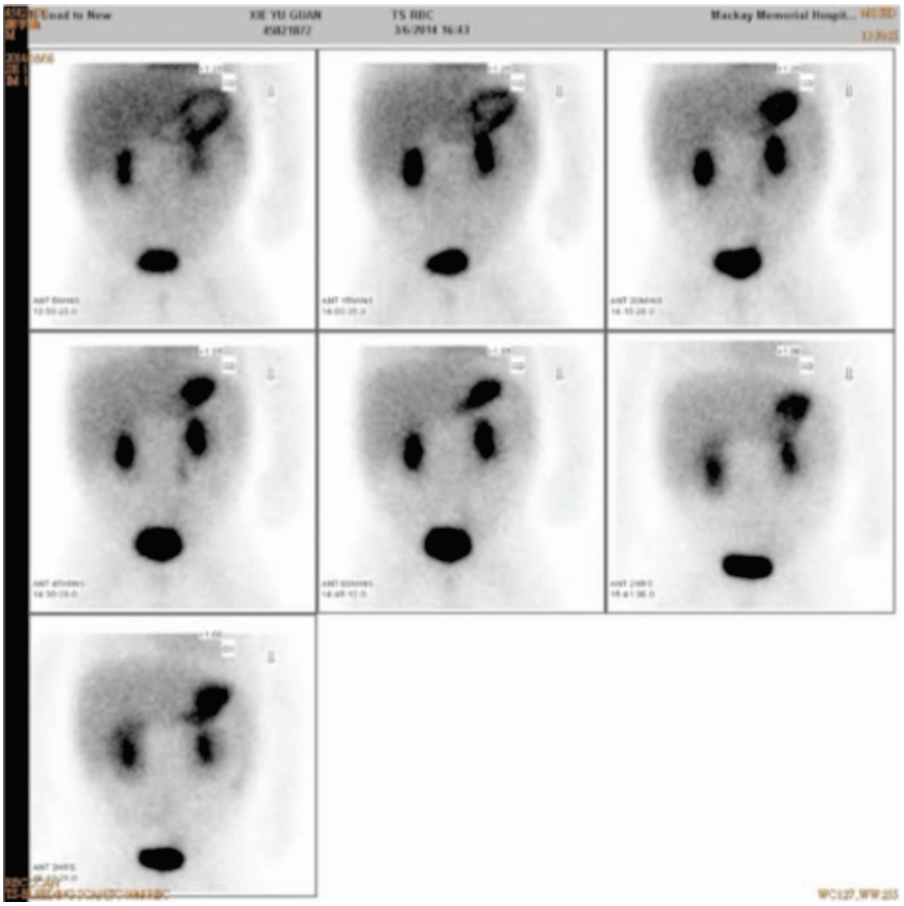

**Figure 3.** Technetium-99m-labeled RBC bleeding scan: the possibility of gastrointestinal tract bleeding in the jejunum.

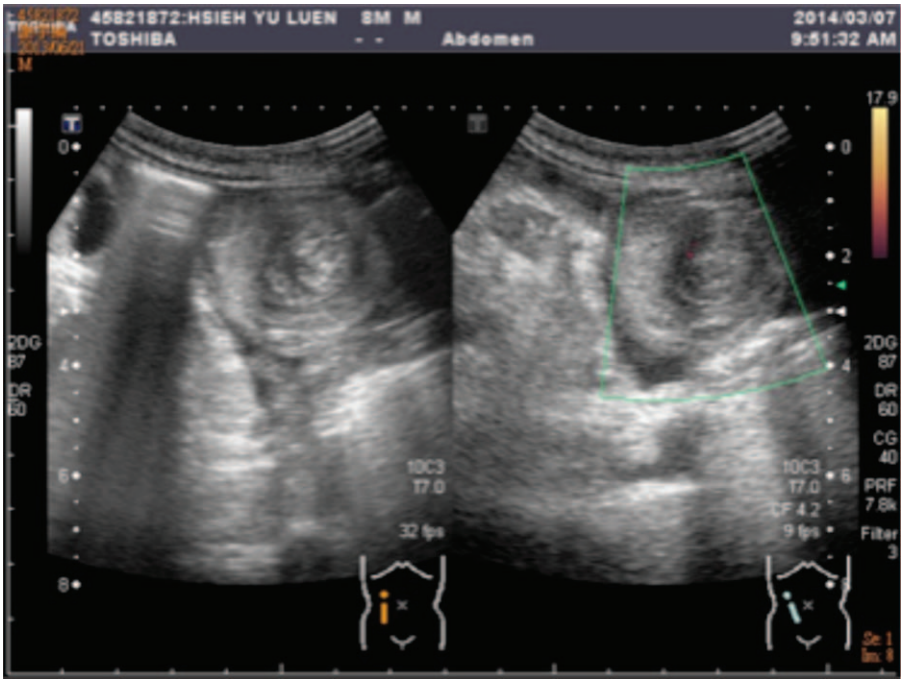


**Figure 4.** Abdominal sonography: multiple target lesions, indicating intussusceptions.

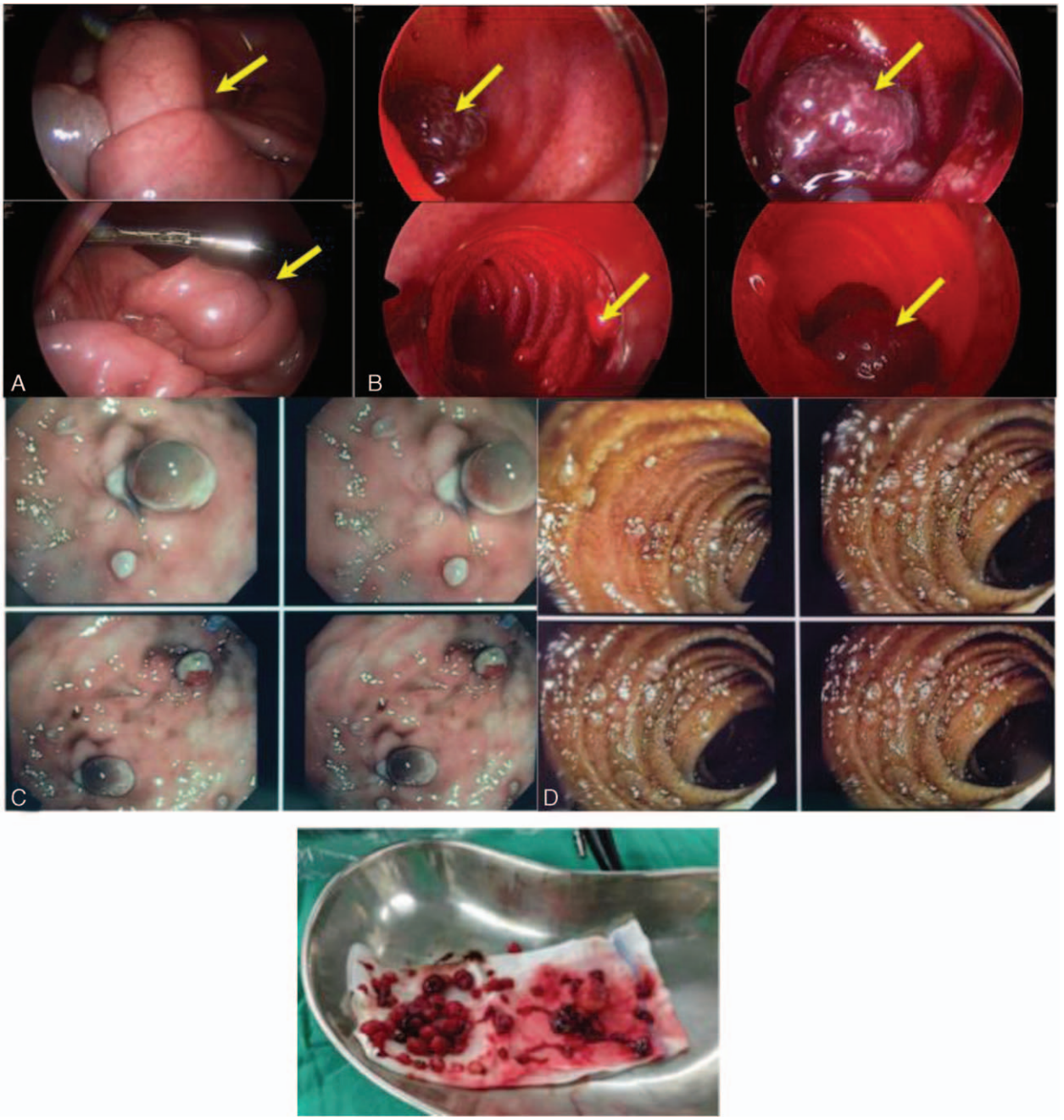


**Figure 5.** -1 Polypectomy and reduction of intussusceptions: (A) 2 intussusceptions were found, 1 at 20 cm distal to Treitz ligament and another 1 at jejunoileal junction. (B, D) Numerous polyps were found in whole small bowel, especially in jejunum. (C) Panendoscopy was done and polyps in stomach and duodenum were found. Figure 5-2—Polypectomy: large polyps were removed.

Thereafter, the patient suffered from multiple episodes of anemia, gastrointestinal hemorrhage, rectal prolapsed polyps, symptomatic colic-colic intussusceptions requiring radiologic reductions, protein-losing enteropathy, and immunodeficiency. It was our impression that the immunodeficiency was secondary to the remarkable protein-losing enteropathy and malnutrition. Human immunodeficiency virus (HIV) infection was excluded due to negative maternal HIV testing during prenatal checkups. It is not a routine in our institution to conduct another HIV examination before 24 months of age in infants with prior negative virologic tests. The serology study showed notably hypoglobulinemia with IgG of 115 mg/dL and IgM of 33 mg/dL. Complement levels were also significantly below the normal limits with C3 of 51 mg/dL and C4 of 7 mg/dL. Intravenous immunoglobulin was administered. In 6 months, polyps recurred and occupied the majority of the gastrointestinal tract. Supportive management was given. The patient expired for severe sepsis at the age of 18 months.

## 3. Discussion

Juvenile polyps are mostly solitary, influencing approximately 1% of preschool and school-aged children.[4] Polyps can be effectively treated with polypectomy, leaving minimal consequences. Juvenile polyposis syndrome may extensively affect a large portion of alimentary tract, and be usually related to

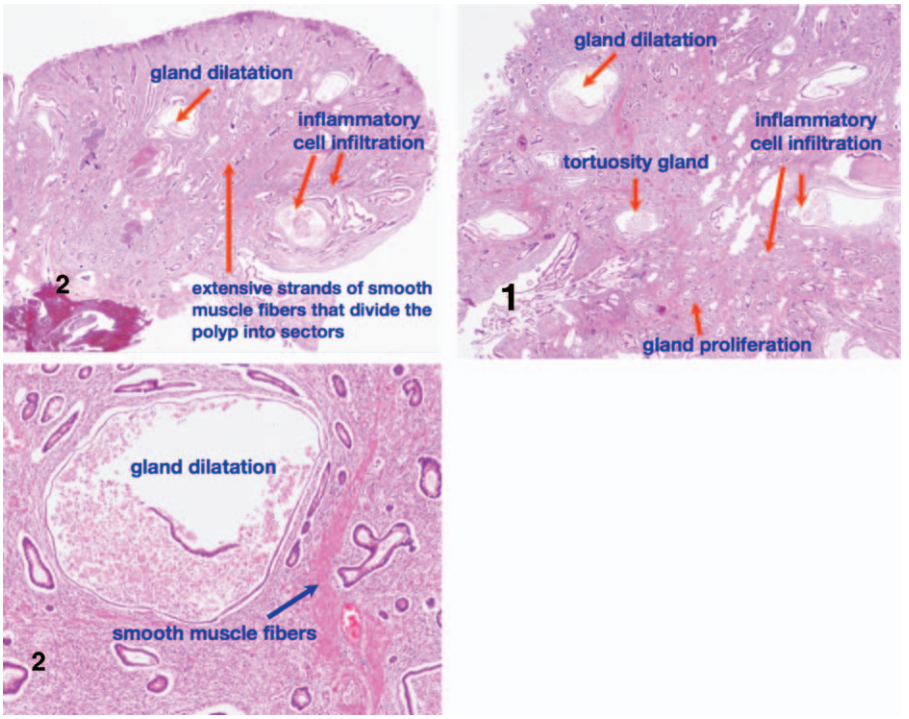


**Figure 6.** Pathology of jejunum: 1. Microscopically, hamartomatous polyps composed of elongation, tortuosity, and dilatation of the gastric foveolae and intestinal mucosal glands. 2. The underlying stroma is characterized by broadband smooth muscle fibers, intermingling with the glands.

malignant potential. It has been known with 3 subtypes: diffuse juvenile polyposis of infancy (<6 months of age), diffuse juvenile polyposis (6 months–5 years of age), and juvenile polyposis coli (5–15 years of age).[1]

Histologically, the polyps in juvenile polyposis syndrome are composed of mucous filled, dilated glands that are often associated with inflammatory cell infiltration. Unlike those in Peutz-Jeghers syndrome, smooth muscle proliferation is rarely seen.[5,6] Nevertheless, the polyps consisted of smooth muscle bands in stroma in this case. Peutz-Jeghers syndrome was initially suspected. However, the patient had neither relevant family history nor oral pigmented lesions that were highly associated with Peutz-Jeghers syndrome.[7] Peutz-Jeghers syndrome was excluded, whereas the diagnosis of juvenile polyposis syndrome was established.[8]

Juvenile polyposis of infancy is an extremely rare genetic disorder. It is featured with widespread hamartomatous polyps in the entire gastrointestinal tract in infants <6 months of age. Most patients will have approximately 50 to 100 colorectal polyps.[1] In our case, the initial manifestations of anemia and gastrointestinal bleeding were present at 8 months of age. It is difficult to know whether the polyps occurred <6 months of age. There were numerous polyps with diverse sizes in the gastrointestinal tract. We presumed that this patient should be categorized to juvenile polyposis of infancy.

With regard to treatment, surgery is the mainstay to remove polyps. Endoscopic polypectomy or segmental bowel resection is suggested for juvenile polyposis of infancy.[5,9] However, repeated operations are usually required because recurrence is not uncommon.[10–12] In our case, minimally invasive surgery was accomplished. Polyps recurred so soon in 6 months. Redo enteroscopic polypectomy was technically difficult as well as might bear much higher risks, so it was abandoned. As nearly the entire small bowel is involved, resection of affected bowels was less considered because it would lead to prominent intestinal failure. The decision was made to perform supportive treatment.

To the best of our knowledge, this is the youngest case reported in the literature.

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