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Juvenile Polyposis Syndrome in a Young Male Patient: A Case Report and Review of Literature

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Abstract **Introduction:** Juvenile polyposis syndrome (JPS) is a rare autosomal dominant disorder marked by multiple gastrointestinal polyps. It often presents with symptoms like lower gastrointestinal bleeding, necessitating early diagnosis and treatment. **Case Presentation:** We present a 13-year-old male with a two-month history of rectal bleeding and prolapse. Colonoscopy and genetic testing confirmed JPS, and the patient underwent total proctocolectomy with ileal pouch-anal anastomosis. **Discussion:** JPS involves multiple juvenile polyps, primarily in the colon and rectum, causing symptoms like bleeding, anemia, and prolapse. It is linked to mutations in SMAD4 or BMPR1A genes, increasing cancer risk. Diagnosis is based on clinical and genetic criteria, with treatment ranging from polypectomy to surgery for severe cases, alongside regular surveillance. The patient underwent successful surgery and is under annual monitoring. **Conclusion:** JPS is a rare precancerous condition typically presenting with gastrointestinal bleeding. Early identification is key for surveillance and interventions to reduce cancer risk.

Keywords: *Rectal bleeding, polyp, Juvenile polyposis, rectal prolapse*

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1. Introduction

Juvenile polyposis syndrome (JPS) is a precancerous condition characterized by the presence of multiple hamartomatous juvenile polyps throughout the gastrointestinal (GI) tract. It is a rare autosomal dominant disorder that significantly increases the risk of developing GI cancers, with an estimated prevalence of one per 100,000 live births [1,2,3,4].

JPS arises from germline mutations in either the SMAD4 or BMPR1A genes, both of which play a critical role in the transforming growth factor-beta (TGF-β) signaling pathway [5,6]. Most patients become symptomatic before the age of 20, with the majority presenting with bleeding or anemia due to GI polyps. Rectal bleeding is the most common initial symptom [7,8].

This work has been reported in line with the SCARE criteria [9].

2. Case Presentation

A 13-year-old male presented to our hospital with a two-month history of painless rectal bleeding. The bleeding occurred primarily at the end of defecation. He also reported a protruding mass through his anus of the same duration, which typically appeared during defecation and spontaneously retracted without manipulation. Additionally, the patient experienced fatigue and blurred vision. There was no family history of colorectal cancer or related conditions.

Physical Examination:

The patient was hemodynamically stable, with a pulse rate of 90 beats per minute, respiratory rate of 16 breaths per minute, and blood pressure of 100/85 mmHg. On examination, he had pale conjunctivae, while abdominal findings were unremarkable. Digital rectal examination

revealed normal anal tone, with no palpable masses or blood detected. Physical examination of other body systems was unremarkable.

Laboratory and Endoscopic Evaluation:

A complete blood count revealed a hemoglobin level of 9 g/dL, microcytosis (MCV: 74.5 fL), and a platelet count of 471,000/μL. Coagulation studies were within normal limits (PT: 12 seconds, PTT: 23.7 seconds, INR: 1.07). An upper gastrointestinal endoscopy showed no abnormalities.

Colonoscopy, however, revealed multiple sessile and pedunculated polyps of varying sizes (0.5–4 cm) throughout the rectum, sigmoid colon, transverse colon, and ascending colon (Figure 1). Histopathological examination of biopsy specimens demonstrated polypoid tissue proliferation with dilated glands and marked inflammatory infiltrates in the stroma. No dysplasia was identified (Figure 2).

Diagnosis and Management:

The findings were consistent with juvenile polyposis syndrome (JPS). The patient and family were counseled, and genetic testing confirmed the diagnosis, revealing significant copy number changes in exons 3 and 8 of the BMPR1A gene, duplication of exon 10 of the SMAD4 gene, and multiple exon copy number changes in the PTEN gene.

Given the diagnosis of JPS complicated by chronic blood loss and anemia, the patient underwent total proctocolectomy (TPC) with ileal pouch-anal anastomosis (IPAA) (Figure 3). He is currently on annual follow-up with colonoscopy, and his most recent evaluation showed no recurrence of polyps.

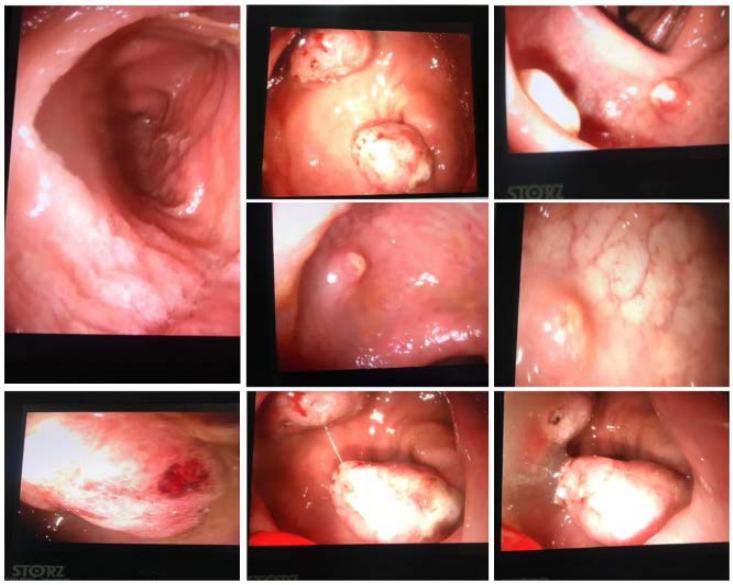

Figure 1. Colonoscopy images: Multiple polyps in different segments of GI tracts

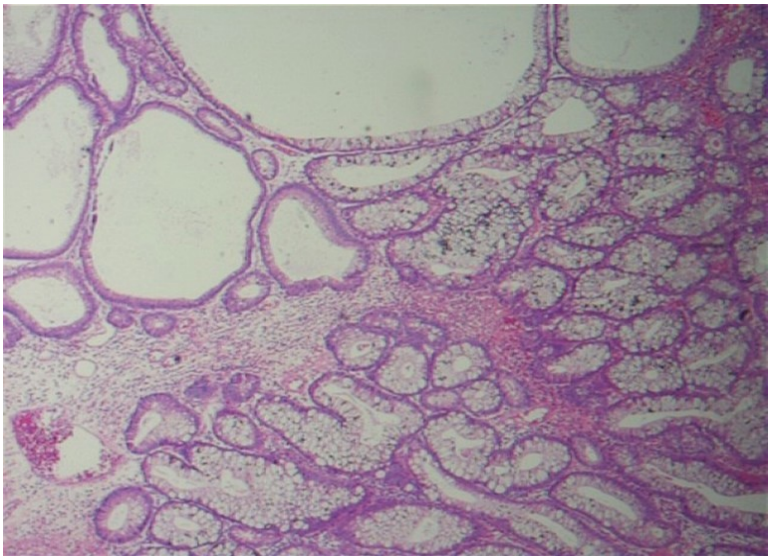

Figure 2. Microscopic examination of polypoid tissue of the colon

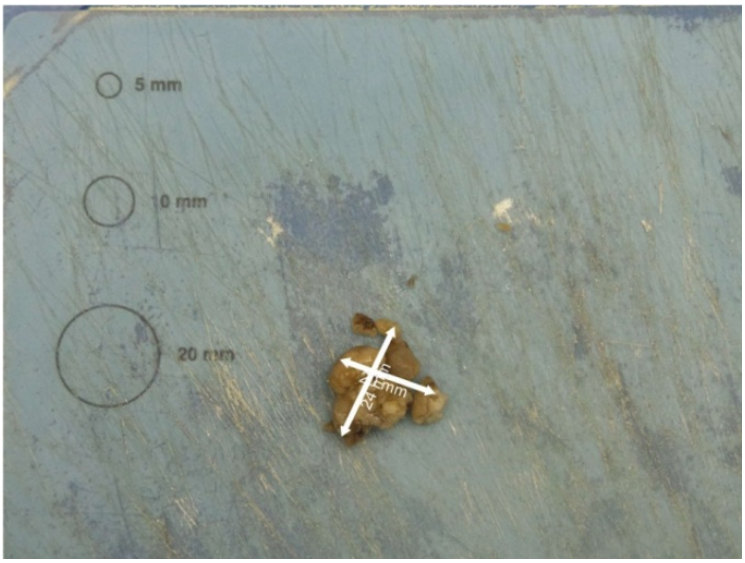

Figure 3. Gross examination of poypoid tissue specimen

3. Discussion

The hallmark clinical feature of JPS is the presence of multiple juvenile polyps in the gastrointestinal (GI) tract, predominantly in the colon and rectum (98%). Polyps may also occur in the stomach (14–28%) and less commonly in the duodenum (7%), jejunum, and ileum (7%) [10].

Most patients present with lower GI bleeding or anemia due to the polyps. Additional symptoms include abdominal pain from intussusception, diarrhea caused by protein-losing enteropathy, and rectal prolapse of polyps [11,12,13,14]. Our patient presented with lower GI bleeding, anemia, and rectal prolapse of a polyp. Endoscopy revealed multiple polyps predominantly in the rectum, sigmoid, transverse, and ascending colon, with no involvement of the upper GI tract.

Mutations in the SMAD4 gene (chromosome 18q21) or the BMPR1A gene (chromosome 10q22) account for 60% of JPS cases. Approximately 25% of cases are sporadic due to de novo mutations, while 75% have a family history of the condition. Both genes are integral to the transforming growth factor-beta (TGF-β) signaling pathway, which regulates cell growth and apoptosis [10].

The clinical diagnosis of JPS can be made when any of the following three criteria are met [15]: a) Five or more juvenile polyps in the colon or rectum, b) Numerous juvenile polyps throughout the GI tract, or c) Any number of juvenile polyps with a family history of JPS.

Patients meeting these criteria should undergo genetic testing for germline mutations in the SMAD4 and BMPR1A genes, which confirm the diagnosis and guide family counseling [16]. Our patient met two diagnostic criteria and underwent genetic testing, which identified mutations in both SMAD4 and BMPR1A, confirming the diagnosis of JPS.

Individuals with JPS face a significantly increased risk of malignancies, particularly colorectal cancer, as well as gastric, duodenal, and pancreatic cancers. The risk arises from adenomatous changes within juvenile polyps. The lifetime risk of colon cancer is estimated at 17–22% by age 35 years and increases to approximately 68% by age 60 [17,18]. Besides cancer, JPS I also associated with several extraintestinal conditions including cardiac (eg, mitral valve prolapse), vascular (eg, arterial aneurysms), skeletal, and cranial abnormalities. Hereditary hemorrhagic telaɴgiectasia (HHT) is seen in around 32% of with *SMAD4* pathogenic variants [2].

Currently, there is no standardized treatment protocol for JPS; management is primarily based on expert opinion and clinical experience.

- **Endoscopic Polypectomy:** This is suitable for small polyps that can be safely removed during endoscopy.
- **Surgical Intervention:** Surgery is recommended in patients with:
  1. Polyps that cannot be removed endoscopically,
  2. Severe blood loss leading to persistent anemia,
  3. Polyps with dysplasia or malignancy, or
  4. A strong family history of colorectal cancer.

Surgical options include colectomy with ileorectal anastomosis (IRA) or total proctocolectomy (TPC) with ileal pouch-anal anastomosis (IPAA). Given our patient's rectal bleeding and persistent anemia, he underwent TPC with IPAA, which successfully addressed his symptoms.

Patients with JPS require regular follow-up and endoscopic surveillance. For individuals with a diagnosis of JPS or a family history of the condition, GI endoscopy should begin at age 12 or earlier if symptoms arise. Surveillance intervals range from 1 to 3 years, depending on polyp burden and progression [11].

Our patient is being monitored with annual colonoscopy and remains in good condition.

4. Conclusion

Juvenile polyposis syndrome is a rare precancerous condition that usually present with lower GI bleeding. Its detection should alert one to consider surveillance, or and intervention to reduce risk of malignancy.

Data sharing statement

Patient's history, physical findings, laboratory investigations, and imaging findings used to support the finding of this study is included in the article**.**

Ethical approval

The author's institution does not require ethical approval for the publication of a single case report.

Consent for publication

Written informed consent was obtained from the patient's parents/legal guardian for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Disclosure

Authors report no conflict of interest.

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