

# Juvenile Polyposis Syndrome Presenting as Intussusception in Adolescent Girl

## Abstract

Juvenile polyposis syndrome is a hereditary condition characterized by the presence of benign hamatomatous polyps in the gastrointestinal tract that typically develop before the age of 20. These polyps can range in number from 5 to 200 and often lead to symptoms such as gastrointestinal bleeding, anaemia, abdominal pain, and diarrhoea. Around 15% of individuals with juvenile polyposis syndrome also have other congenital anomalies, such as cleft palate, polydactyly, and abnormalities in the genital and urinary tracts.

The World Health Organization has established criteria for diagnosing juvenile polyposis syndrome, which include:

1. Having more than 5 polyps in the colon or rectum.
2. Finding juvenile polyps in other parts of the gastrointestinal tract.
3. Identifying juvenile polyps in one or more affected family members. Individuals with generalized juvenile polyposis and juvenile polyposis coli typically develop these polyps during childhood.

It's important to note that most juvenile polyps have the potential to become cancerous, with a 10-50% risk of developing gastrointestinal tract cancer. Colorectal cancer is the most common type of cancer associated with this syndrome.

We are reporting a rare case of juvenile polyposis syndrome in an 18-year-old adolescent girl who presented with ileo-ileal intussusception. She was admitted to our centre as an emergency case due to acute intestinal obstruction caused by this condition. She underwent open surgery, which involved segmental small bowel resection with stapler anastomosis. During the surgery, the entire small bowel was examined, and numerous single polyps were removed through enterotomy and polypectomy.

## Key words

Juvenile polyps, Intussusception, Peutz- Jeghres Syndrome.

## Introduction

Juvenile polyposis syndrome is a relatively rare autosomal dominant disorder characterized by the presence of multiple hamatomatous polyps throughout the gastrointestinal tract. Its estimated incidence is approximately 1 in 100,000 individuals. Those affected by juvenile polyposis syndrome face an increased risk of developing gastrointestinal malignancies, particularly colorectal and gastric cancers, with an estimated risk of about 50% and 20%, respectively. [2,5,]

Similar hamatomatous polyposis syndromes include Peutz-Jeghres syndrome and Cowden syndrome, typically manifesting with benign polyps before the age of 20. Juvenile polyposis syndrome can be inherited in an autosomal dominant manner within families, and it is associated with two specific genes, *BMPRI1A* and *SMAD4*. This means that an alteration in only one copy of the gene is sufficient to develop the condition.

There are three distinct types of juvenile polyposis syndrome:

1. Juvenile polyposis of infancy: This is the most severe form, primarily affecting infants and young children.
2. Generalized juvenile polyposis: This is the most common form, characterized by the presence of polyps in various parts of the gastrointestinal tract, including the stomach, colon, and small intestine.
3. Juvenile polyposis coli: This type of JPS primarily leads to the development of polyps within the colon.

Clinically, individuals with juvenile polyposis syndrome may present with symptoms such as rectal bleeding, diarrhoea, anaemia, and bowel obstruction, often resulting from conditions like intussusception. [1,2,5,7]

### Case Report

On October 5, 2012, an 18-year-old adolescent girl was admitted to our centre due to complaints of abdominal pain, vomiting, and abdominal distension that had persisted for three days. Radiological evaluation revealed significant findings, with an abdominal X-ray indicating distended small bowel loops with air-fluid levels, suggestive of acute intestinal obstruction. Abdominal ultrasonography displayed a distinctive "Target" or "Doughnut" sign in a transverse plane, further supporting the diagnosis. An abdominal CT scan confirmed the presence of small bowel dilation and identified a sausage-shaped mass, characteristic of ileo-ileal intussusception.

The diagnosis of intestinal obstruction secondary to ileo-ileal intussusception was established, prompting immediate action. The patient was taken to the operating room for open surgery. During the procedure, manual reduction of the ileo-ileal intussusception was successfully performed. While palpating the small bowel, an intraluminal mass was discovered. Consequently, a segmental resection of the small intestine was conducted, followed by stapler anastomosis. Surprisingly, upon thorough examination of the entire small bowel with manual palpation, multiple polyps were identified in the distal segment. To address this unexpected finding, multiple enterotomies were performed, along with the necessary polypectomies. Following an uneventful recovery period, the patient was discharged on the 8th day post-surgery.

Gross pathology examination of the specimen revealed multiple polypoid masses resembling grapes. These polyps were pedunculated and measured between 1 to 2 cm in diameter. The mass exhibited classic characteristics of hamatomatous polyps. Subsequent histopathological analysis confirmed the diagnosis of juvenile polyposis syndrome. There have been no recurrences in the five years since the follow-up. (Fig 1-12)



Fig-1 USG abdomen showing "Target Sign" or "Doughnut Sign"

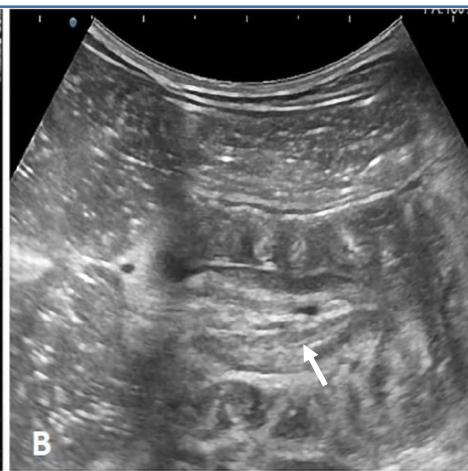
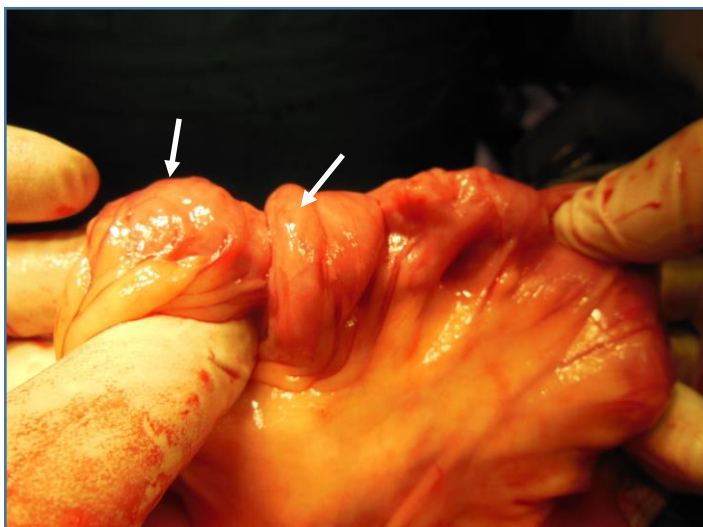


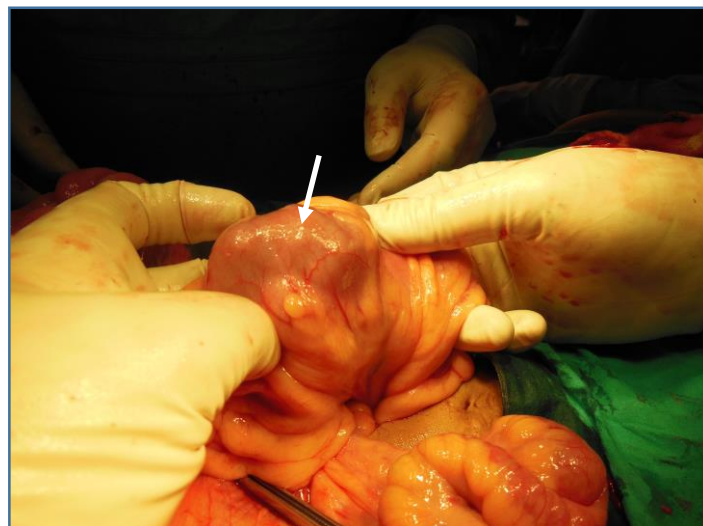
Fig-2 USG abdomen showing "Sandwich Sign" or "Pseudo-kidney Sign"



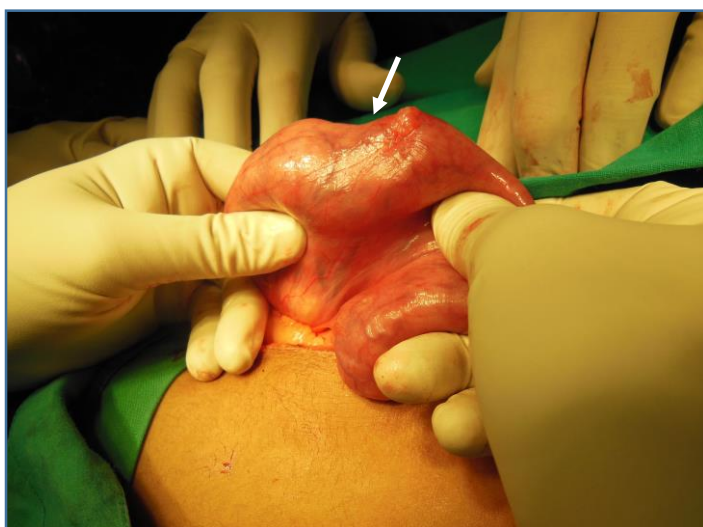
Fig-3 CT abdomen showing "Target Sign" or "Sausage Sign"



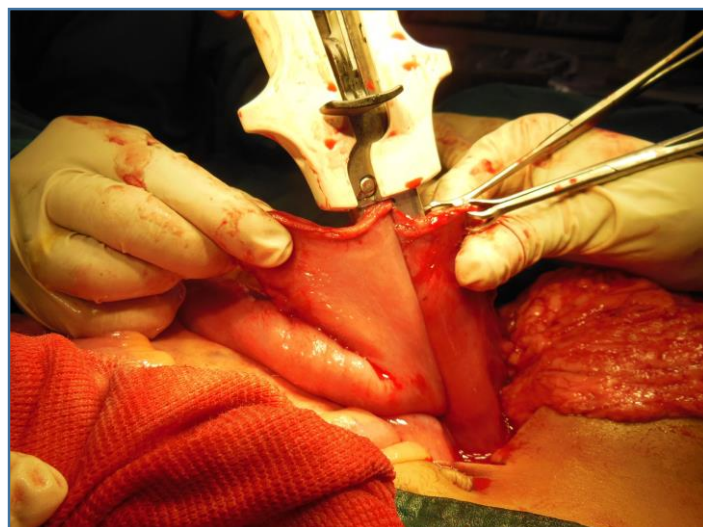
**Fig-4 Intraoperative photographs showing ileo-ileal intussusception**



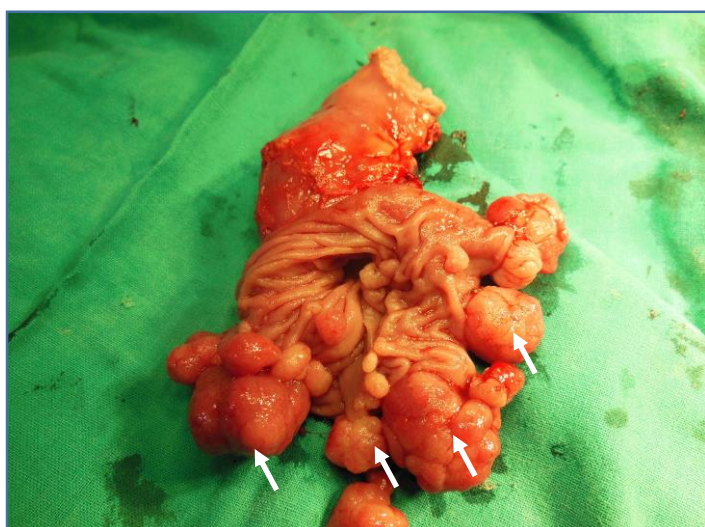
**Fig-5 Intraoperative photographs showing ileo-ileal intussusception with internal mass**



**Fig-6 Intraoperative photographs showing ileo-ileal intussusception with internal mass**



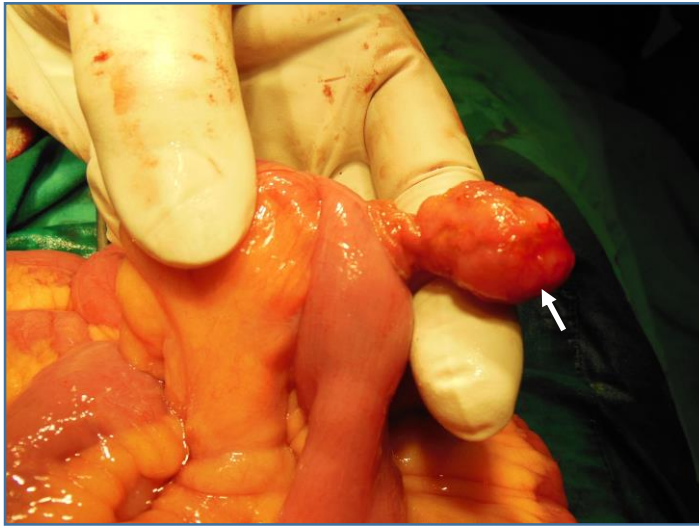
**Fig-7 Intraoperative photographs showing ileo-ileal Stapler anastomosis**



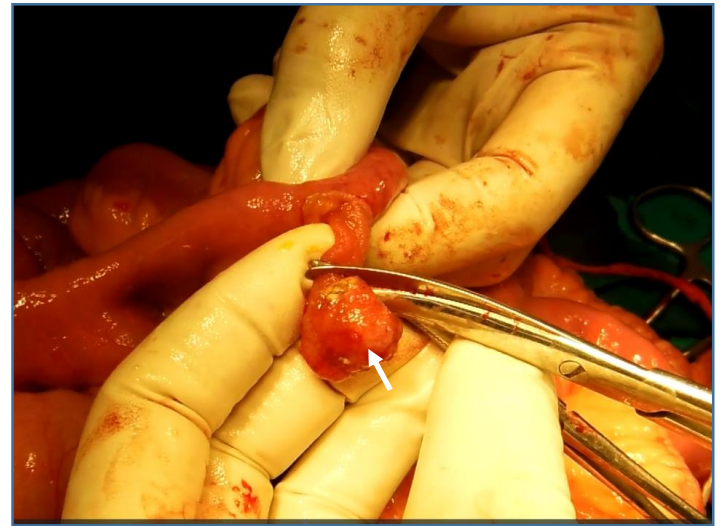
**Fig-8 photographs showing Multiple grape like polyps**



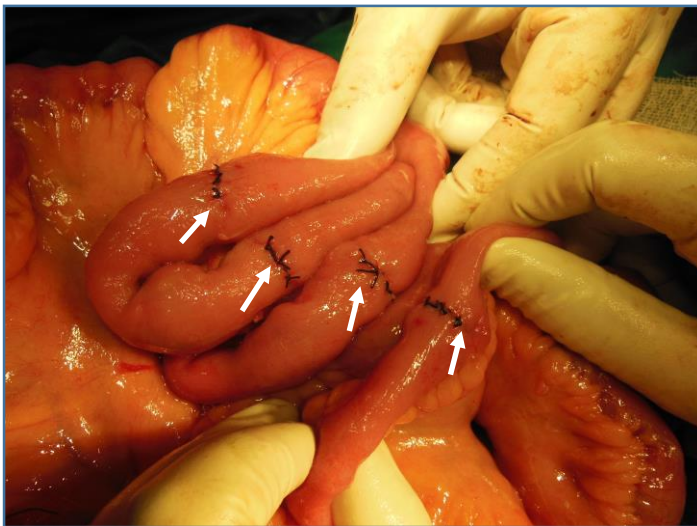
**Fig-9 photographs showing Intraluminal multiple grape like polyps**



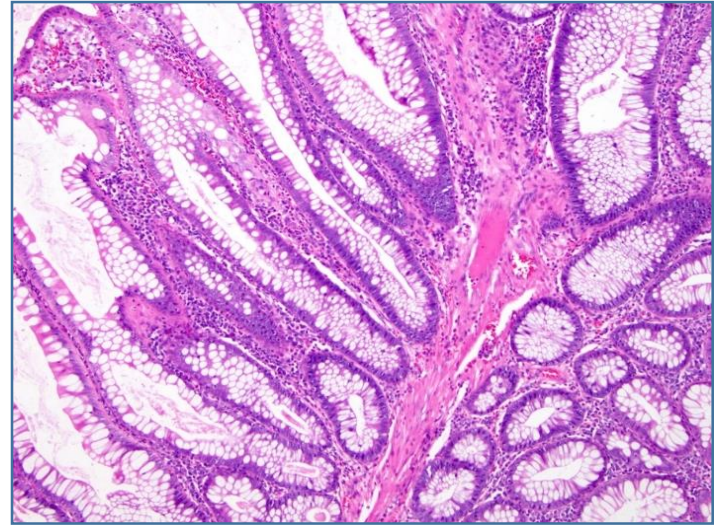
**Fig-10 Intraoperative photographs showing a single polyp after enterotomy**



**Fig-11 Intraoperative photographs showing excision of single polyp**



**Fig-12 Intraoperative photographs showing multiple enterotomy incisions after polypectomies**



**Fig-13 Microscopic features consistent with Juvenile polyp**

## Discussion

The term "intussusception" was first coined by John Hunter in 1789 to describe a condition in which one segment of the intestine telescopes into another, causing a blockage. In paediatric cases, intussusception is most often idiopathic and occurs in the ileocolic region. The most common triggers include Meckel's diverticulum, duplication cysts in the small bowel, and juvenile polyps. The initial diagnostic approach typically involves abdominal ultrasonography, which can reveal characteristic findings such as the "Target" or "Doughnut Sign" in the transverse plane and the "Pseudo-kidney" or "Hayfork Sign" in the longitudinal plane. [3,5,7,]

For a more comprehensive evaluation, an abdominal CT scan is often employed, with a diagnostic accuracy ranging from 60% to 100%. Confirming the diagnosis of juvenile polyposis syndrome requires diagnostic tests like upper GI endoscopy, colonoscopy, and genetic blood testing. Mutations in the BMPRIA and SMAD4 genes are responsible for causing juvenile polyposis syndrome. [4,5]

Juvenile polyposis syndrome follows an autosomal dominant inheritance pattern, with roughly 75% of cases stemming from inheriting the mutation from an affected parent. The remaining 25% of

cases arise from new mutations in individuals without a family history of the disorder. In families affected by juvenile polyposis syndrome, the lifetime risk of developing GI cancer varies from 11% to 86%. Colorectal cancer is reported in 17-68% of cases by the age of 60, and there is a 21% incidence of gastric cancer. When intussusception is diagnosed, the next step involves attempting reduction through a radiological procedure using either a liquid contrast enema or an air contrast enema. [5,7]

### **Treatment**

In adult cases of intussusception, the preferred treatment approach is open laparotomy and surgical resection of the affected bowel. En-block resection, with or without reduction of the involved bowel, is commonly recommended to prevent bowel necrosis, perforation, and the potential spread of cancer cells to other areas. Surgical options may include polypectomy or enterectomy with ileo-ileal anastomosis. Surgical intervention remains the primary choice for managing adult intussusception. [5,7]

In recent years, there has been a growing adoption of laparoscopic techniques for the treatment of adult intussusception. With the laparoscopic approach, it is possible to successfully perform intracorporeal reduction of intussusception and segmental resection of the small bowel. Therefore, in cases of small bowel intussusception in adults, surgeons may consider using a laparoscopic approach as the initial treatment option. [4,5,]

Juvenile polyposis syndrome (JPS) currently has no cure. Early detection and prompt treatment of polyps are crucial for achieving the best outcomes. Individuals diagnosed with JPS face a 30% to 50% lifetime risk of developing GI tract cancer. To mitigate this risk, it is recommended to conduct annual screenings using colonoscopy, upper GI endoscopy should also be repeated every 1 to 3 years, depending on the burden or number of polyps, starting at the age of 12 to 15 years. Regular monitoring and intervention can help manage the risk associated with JPS. [3,5,7]

### **Conclusion**

Juvenile polyposis syndrome is a rare genetic disorder characterized by the presence of numerous distinctive juvenile polyps in the gastrointestinal tract. It is inherited in an autosomal dominant manner. Intussusception, on the other hand, is a surgical emergency necessitating surgical intervention.

### **Reference**

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