Casestudy

Juvenile PolyposisSyndromePresentingasIntussusceptionin AdolescentGirl

Abstract

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

We are reporting a rare case of juvenile polyposis syndrome in an 18-year-old adolescent girl whopresented with ileo-ileal intussusception. She was admitted to our centre as an emergency case due toacute intestinal obstruction caused by this condition. She underwent open surgery, which involvedsegmental small bowel resection with stapler anastomosis. During the surgery, the entire small bowelwasexamined,andnumeroussinglepolypswereremovedthroughenterotomyandpolypectomy.

Keywords

Juvenile polyposis, polyps, adolescents, benign nodules, syndrome, gastrointestinal problems, intestinal obstruction, abdominal pain, autosomal dominant inheritance, genetic mutation.

Introduction

Juvenile polyposis syndrome is a relatively rare autosomal dominant disorder characterized by thepresence of multiple hamatomatous polyps throughout the gastrointestinal tract. Its estimatedincidence is approximately 1 in 100,000 individuals. Those affected by juvenile polyposis syndromeface an increased risk of developing gastrointestinal malignancies, particularly colorectal and gastriccancers,withan estimated riskofabout50%and 20%,respectively.[2,5,]

Similar hamatomatous polyposis syndromes include Peutz-Jeghres syndrome and Cowdensyndrome, typically manifesting with benign polyps before the age of 20. Juvenile polyposissyndrome can be inherited in an autosomal dominant manner within families, and it is associated withtwo specific genes, BMPR1A and SMAD4. This means that an alteration in only one copy of the geneissufficientto developthecondition.

Therearethreedistincttypesofjuvenilepolyposissyndrome:

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

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

CaseReport

On October 5, 2012, an 18-year-old adolescent girl was admitted to our centre due to complaintsof abdominal pain, vomiting, and abdominal distension that had persisted for three days. Radiologicalevaluation revealed significant findings, with an abdominal X-ray indicating distended small bowelloops with air-fluid levels, suggestive of acute intestinal obstruction. Abdominal ultrasonographydisplayed a distinctive "Target" or "Doughnut" sign in a transverse plane, further supporting thediagnosis. An abdominal CT scan confirmed the presence of small bowel dilation and identified asausage-shapedmass, characteristic ofileo-ilealintussusception.

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. Duringthe procedure, manual reduction of the ileo-ileal intussusception was successfully performed. Whilepalpating the small bowel, an intraluminal mass was discovered. Consequently, a segmental resectionof the small intestine was conducted, followed by stapler anastomosis. Surprisingly, upon thoroughexamination of the entire small bowel with manual palpation, multiple polyps were identified in thedistal segment. To address this unexpected finding, multiple enterotomies were performed, along withthe necessary polypectomies. Following an uneventful recovery period, the patient was discharged onthe8th daypost-surgery.

Gross pathology examination of the specimen revealed multiple polypoid masses resemblinggrapes. These polyps were pedunculated and measured between 1 to 2 cm in diameter. The massexhibited classic characteristics of hamatomatous polyps. Subsequent histopathological analysisconfirmed the diagnosis of juvenile polyposis syndrome. There have been no recurrences in the fiveyearssincethefollow-up. **(Fig 1-12)**

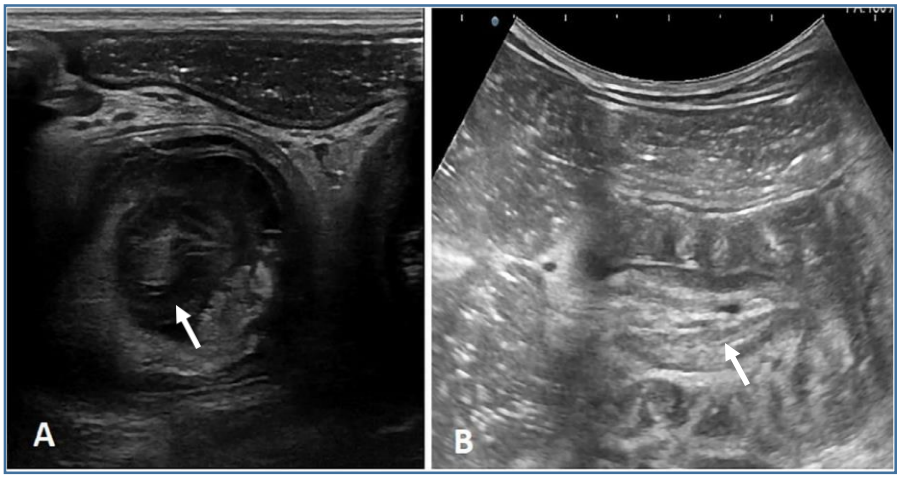

Fig-1 USG abdomen showing“TargetSign”or“DoughnutSign”

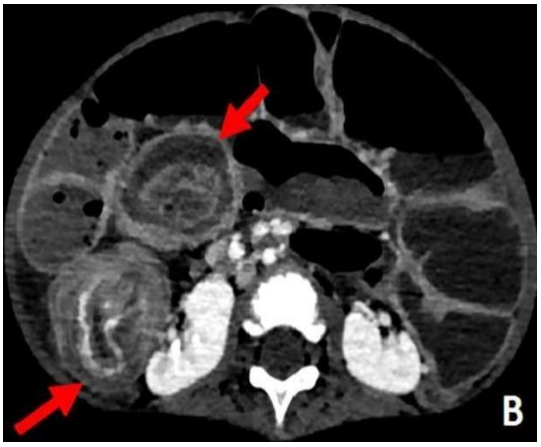

Fig-2USGabdomenshowing“SandwichSign”or“Pseudo-kidneySign”

Fig-3CTabdomenshowing“TargetSign”or“Sausage Sign”

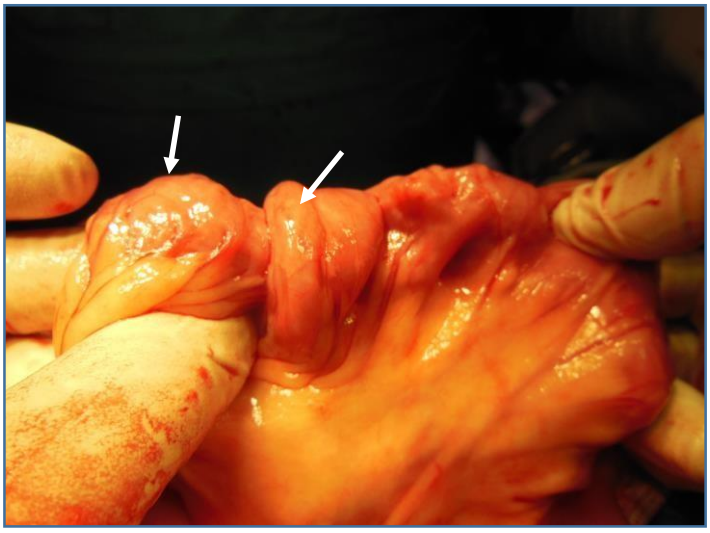

Fig-4Intraoperativephotographsshowingileo-ilealintussusception

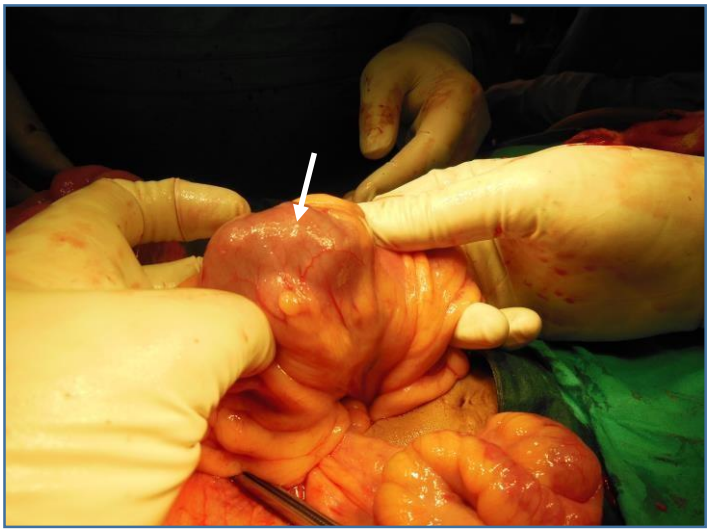

Fig-5 Intraoperative photographs showingileo-ilealintussusceptionwithinternalmass

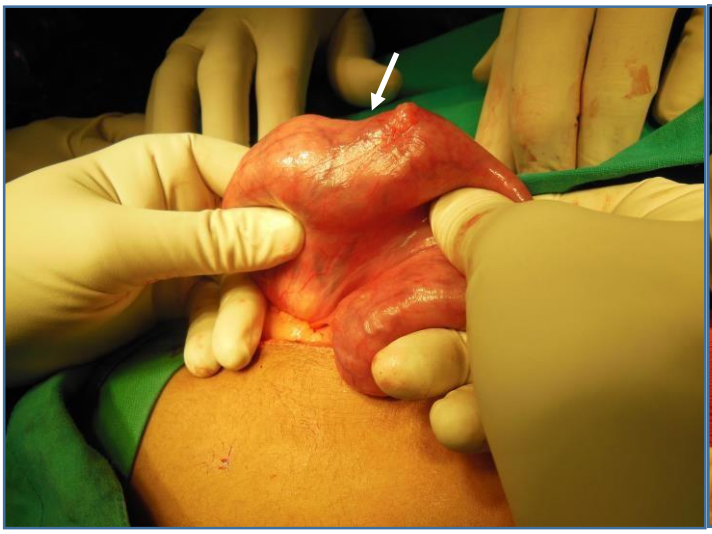

Fig-6 Intraoperative photographs showingileo-ilealintussusceptionwithinternalmass

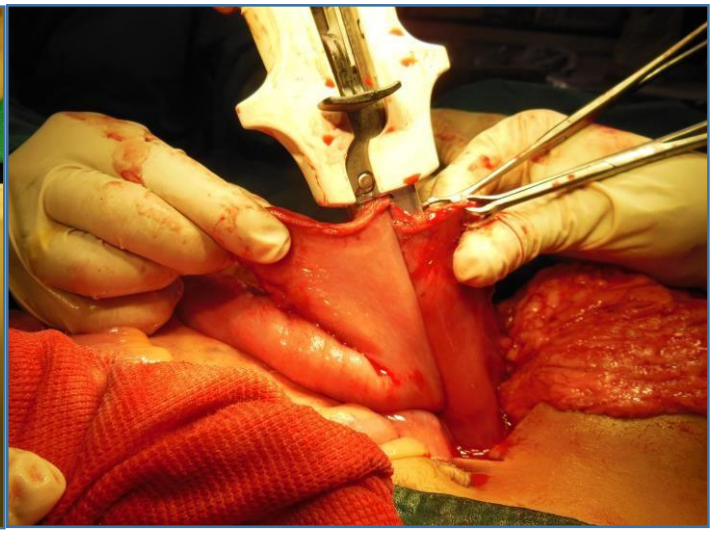

Fig-7 Intraoperative photographs showingIleo-ileal Stapleranastomosis

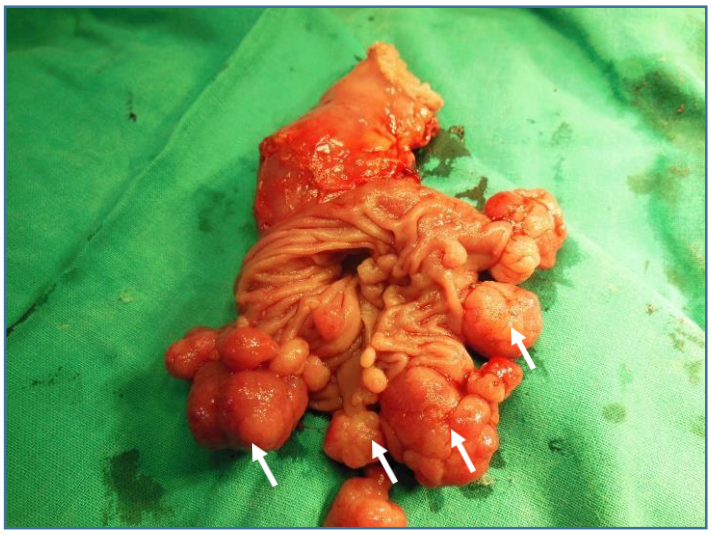

Fig-8 photographs showingMultiplegrapelike polyps

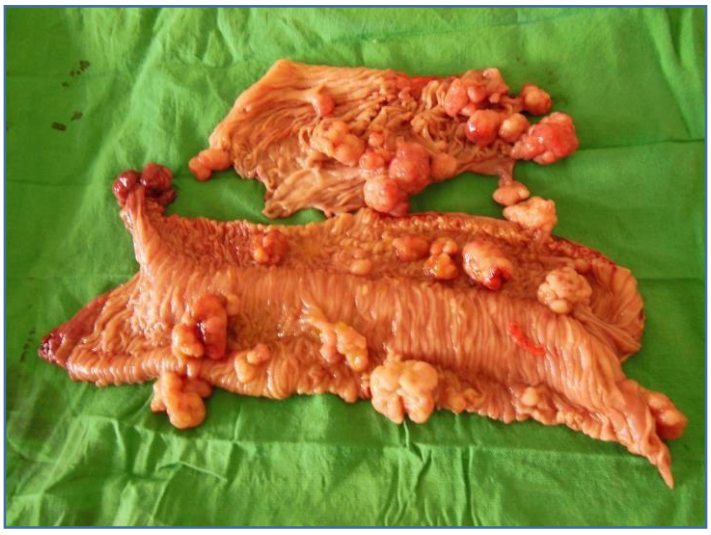

Fig-9 photographs showingIntraluminalmultiplegrapelike polyps

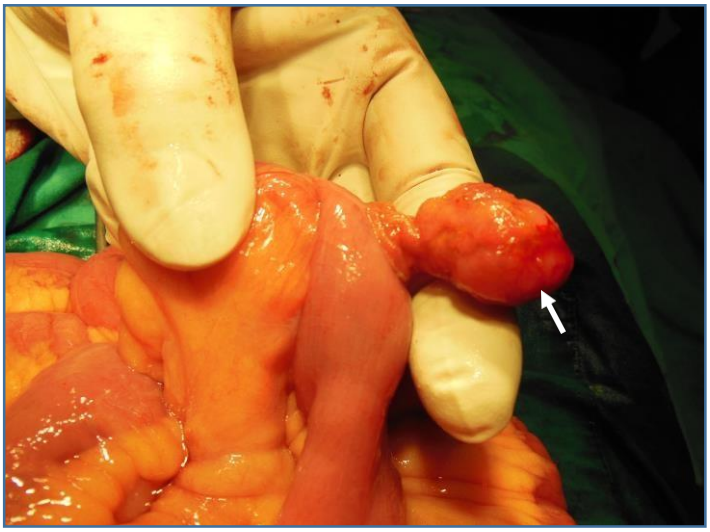

Fig-10Intraoperativephotographsshowingasinglepolypafterenterotomy

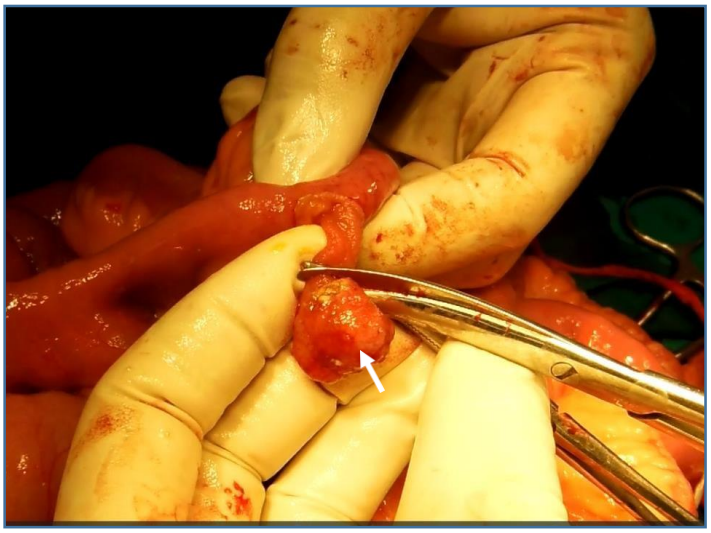

Fig-11 Intraoperative photographs showingexcisionofsingle polyp

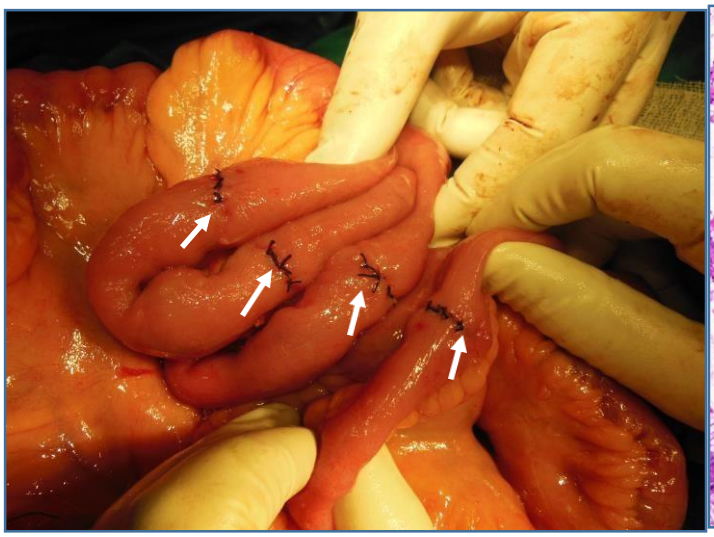

Fig-12 Intraoperative photographs showingmultipleenterotomyincisionsafterpolypectomies

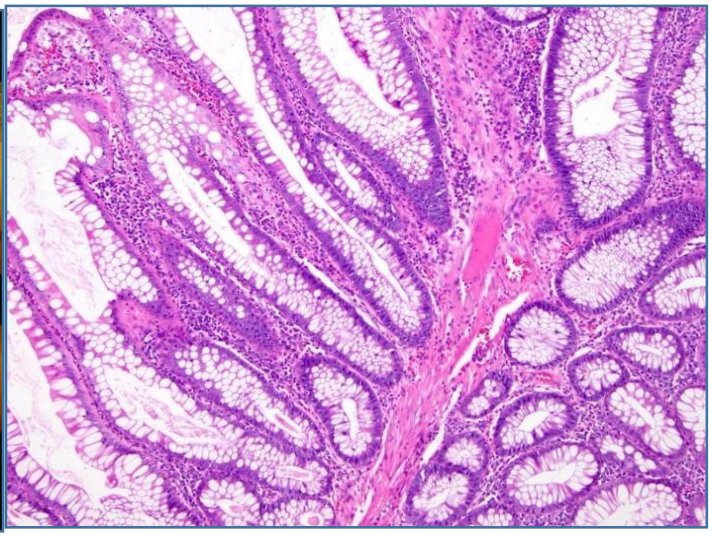

Fig-13 Microscopic features consistent withJuvenilepolyp

Discussion

The term "intussusception" was first coined by John Hunter in 1789 to describe a condition inwhich 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 commontriggers include Meckel's diverticulum, duplication cysts in the small bowel, and juvenile polyps. Theinitial diagnostic approach typically involves abdominal ultrasonography, which can revealcharacteristic findings such as the "Target" or "Doughnut Sign" in the transverse plane and the"Pseudo-kidney"or "Hayfork Sign" inthelongitudinalplane.[3,5,7,]

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

1. Havingmorethan5polypsinthecolonorrectum.
2. Findingjuvenilepolypsinotherpartsofthegastrointestinaltract.
3. Identifying juvenile polyps in one or more affected family members. Individuals withgeneralized juvenile polyposis and juvenile polyposis coli typically develop these polypsduringchildhood.

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 ofcancerassociated withthissyndrome.[1,23]

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

Juvenile polyposis syndrome follows an autosomal dominant inheritance pattern, with roughly75%of casesstemmingfrominheritingthe mutationfroman affectedparent.Theremaining25%of

cases arisefromnewmutationsinindividualswithouta familyhistoryofthedisorder.Infamiliesaffected by juvenile polyposis syndrome, the lifetime risk of developing GI cancer varies from 11% to86%. Colorectal cancer is reported in 17-68% of cases by the age of 60, and there is a 21% incidenceof gastric cancer. When intussusception is diagnosed, the next step involves attempting reductionthrougharadiologicalprocedureusingeitheraliquidcontrastenemaoranaircontrast enema.[5,7]

Treatment

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

In recent years, there has been a growing adoption of laparoscopic techniques for the treatment ofadult intussusception. With the laparoscopic approach, it is possible to successfully performintracorporeal reduction of intussusception and segmental resection of the small bowel. Therefore, incases of small bowel intussusception in adults, surgeons may consider using a laparoscopic approachasthe initialtreatmentoption.[4,5,]

Juvenile polyposis syndrome (JPS) currently has no cure. Early detection and prompt treatment ofpolyps 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 annualscreenings using colonoscopy, upper GI endoscopy should also be repeated every 1 to 3 years,dependingontheburdenornumberofpolyps,startingattheageof12to15years.Regularmonitoringandi ntervention canhelp manage theriskassociated withJPS.[3,5,7]

Conclusion

Juvenile polyposis syndrome is a rare genetic disorder characterized by the presence of numerousdistinctive juvenile polyps in the gastrointestinal tract. It is inherited in an autosomal dominantmanner. Intussusception, on the other hand, is a surgical emergency necessitating surgicalintervention.

Consent

As per international standards or university standards, patients' written consent has been collected and preserved by the author(s).

Ethical Approval:

As per international standards or university standards guideline participant consent and ethical approval has been collected and preserved by the authors.

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