Juvenile Polyposis Syndrome in a 16-year-old Girl: a Case Report From a Resource-Limited Setting

[1]MA Ituen, [1]EI Akpanudo, [2]CS Eyo

Abstract

Background: Juvenile Polyposis Syndrome (JPS) is a rare autosomal dominant disorder characterised by multiple hamartomatous polyps in the gastrointestinal tract. It can lead to complications such as gastrointestinal bleeding, anaemia, bowel obstruction, and an increased risk of malignancy. Management typically requires early diagnosis, genetic evaluation, surgical intervention, and long-term surveillance.

Aim/Objective: To report a case of Juvenile Polyposis Syndrome in a child and document the challenges of diagnosis and management in a resource-limited setting.

Case Report: A 16-year-old girl presented with an 11-year history of rectal bleeding and a 3-year history of a prolapsing rectal mass. She was severely underweight, anaemic, and prepubertal. Examination revealed multiple friable, pedunculated polyps prolapsing from the rectum. A double-contrast barium enema demonstrated diffuse colorectal involvement. In the absence of colonoscopy and genetic testing, surgical planning was guided by intraoperative findings. A total proctocolectomy with temporary ileostomy was performed, followed by ileoanal anastomosis nine months later. Histology confirmed multiple hamartomatous polyps with low-grade dysplasia.

Conclusion: This case highlights the challenges of managing JPS in resource-constrained settings like ours, where the limited diagnostic tools and financial constraints can delay diagnosis and hinder long-term care. Improving awareness and access to appropriate diagnostic and surgical services is key to better outcomes.

Keywords: Juvenile Polyposis Syndrome, Hamartomatous polyps, Colorectal Neoplasms, Total proctocolectomy, Resource-limited setting.

Nigerian Research J Clin. Sci. 2025; 15(1): 13-19

[1]Paediatric Surgery Unit, Department of Surgery, University of Uyo, Nigeria
[2]Department of Anaesthesia, University of Uyo, Nigeria

Corresponding Author: Dr. Monday Akpabio Ituen. Paediatric Surgery Unit, Department of Surgery, Faculty of Clinical Sciences University of Uyo, Nigeria. Email: ima.ituen@yahoo.com

Introduction

Gastrointestinal polyps, which can occur anywhere in the alimentary tract of a child, are commonly benign and isolated lesions.[1] However, some children with juvenile polyps may have hereditary syndromes that predispose them to adenomatous or hamartomatous polyposis. The hereditary polyposis syndromes include adenomatous syndromes like Familial Adenomatous Polyposis and Gardner syndrome, as well as hamartomatous polyposis syndromes such as Peutz-Jeghers syndrome, Juvenile polyposis syndrome, and Cowden's disease. Hamartomatous polyps are the most common type found in children.[2]

Juvenile polyposis syndrome (JPS) is a rare autosomal dominant disorder in which the patient develops multiple polyps predominantly in the stomach and/or the colon. It is estimated to occur in approximately 1 in 100,000 to 1 in 160,000 individuals in the population.[3] The main concerns with JPS are the risk of gastrointestinal complications, such as bleeding, obstruction, infarction, malnutrition, and intussusception, which may lead to significant morbidity and mortality, and the increased risk of malignancy. The lifetime risk of gastrointestinal cancer in JPS ranges from 11 to 86%.[4]

Managing hereditary polyposis syndromes like JPS in resource-limited settings is challenging due to limited access to essential diagnostic tools, such as paediatric endoscopy and genetic testing. This often results in delayed diagnosis and suboptimal treatment, especially for patients with prolonged illness. In this report, we describe the case of an adolescent female with colorectal polyposis managed in a resource-limited setting.

Case presentation

A 16-year-old girl presented to the outpatient clinic with an 11-year history of rectal bleeding and a 3-year history of grape-like mass protruding from her anus. Initially, the mass reduced spontaneously, but over the last 12 months, it required manual reduction. The rectal bleeding was profuse, necessitating multiple blood transfusions within the last year. She was the firstborn child in her family, and there were no other first-degree relatives with similar symptoms.

On physical examination, she was pale with no jaundice or oedema. Oral examination showed no hyperpigmentation of her buccal mucosa. She was afebrile with stable vital signs. She weighed 31.6 kg (< 5th percentile; 59% ideal body weight). Her breast development and pubic hair were both at Tanner Stage 1 (prepubertal).

Her abdomen was not distended. It was soft on palpation with no areas of tenderness, and bowel sounds were normal. Digital rectal examination revealed a normal perianal region with good anal sphincteric tone. Several soft polypoid masses were palpated in the rectum, extending to the anal canal. The examining finger was blood stained. On gentle straining, a large bleeding mass prolapsed, containing multiple pedunculated polyps that were larger than 1 cm and friable (Figure 1).

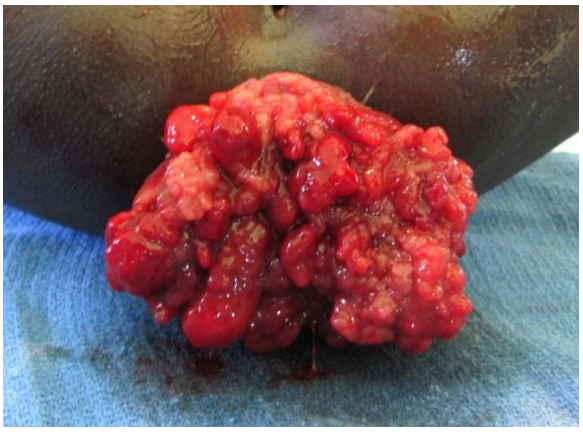

Figure 1: Prolapsed rectal mass with multiple pedunculated, bleeding polyps.

Her haematocrit was 23%, haemoglobin 7.7 g/dl, and her total white cell count was 15.7 x 10^9/L with 72% neutrophilia. Other laboratory tests were within normal ranges, and an abdominal ultrasound scan was normal. A double-contrast barium enema demonstrated multiple round filling defects diffusely scattered throughout the large bowel, extending to the rectum, consistent with widespread colorectal involvement. However, there were no facilities for a paediatric endoscopy and colonoscopy to assess her entire gastrointestinal tract, nor was genetic testing available.

We discussed with the patient and her family the need for large bowel resection to stop the ongoing bleeding and reduce the risk for malignant transformation. She was optimized and subsequently had an exploratory laparotomy, which revealed an unremarkable peritoneal cavity with no evidence of tumour seeding. Multiple polypoid masses were palpated along the entire colon from the caecum to the rectum. The terminal ileum was normal on palpation. The intraoperative decision was to offer the patient a staged reconstruction. Proctocolectomy and temporary end ileostomy was carried out in the first stage. The resected colonic specimen showed multiple pedunculated polyps (Figure 2). Histology revealed multiple hamartomatous polyps with multiple foci of low-grade dysplasia. Following nutritional rehabilitation as an outpatient, she underwent ileoanal anastomosis nine months later. Her postoperative course was uneventful, and she was discharged three weeks after surgery. Over the next ten months, she was followed up in the outpatient clinic, during which time she showed considerable weight gain and reported opening her bowels up to three times a day, which was acceptable to her. The patient and her family were counselled on the need for life-long surveillance, and her family was advised on the need to screen other family members. Unfortunately, she was subsequently lost to follow-up after 10 months.

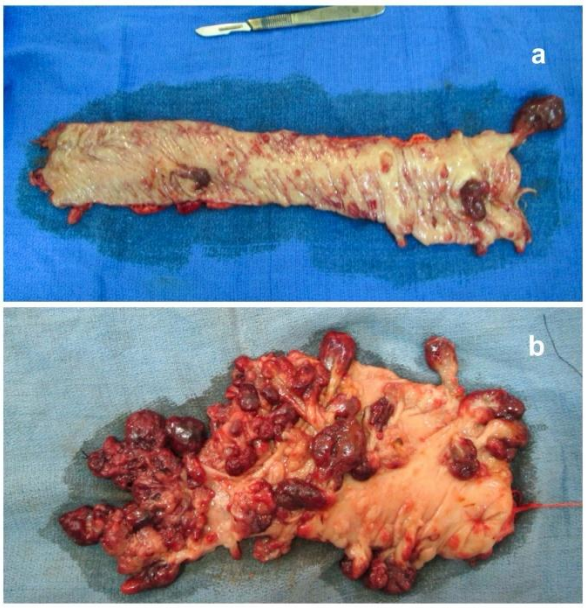

Figure 2 (a) A segment of the resected colon showing multiple mucosal polyps. (b) Proctocolectomy specimen displaying

numerous pedunculated polyps involving the rectum and distal colon.

Discussion

Juvenile Polyposis Syndrome (JPS) is characterized by a predisposition to hamartomatous polyps in the gastrointestinal (GI) tract, specifically in the stomach, small intestine, colon, and rectum. It is estimated that 40–60% of JPS cases are caused by mutations in the BMPR1A and SMAD4 genes (located on chromosome 10q23.2 and 18q21.1 respectively), both linked to the TGF-B/BMP signaling pathway.[5] Individuals with SMAD4 mutations often have a more severe phenotype, frequently associated with a combined syndrome known as hereditary hemorrhagic telangiectasia (HHT), which involves vascular malformations that can lead to life-threatening bleeding episodes (such as epistaxis, mucocutaneous telangiectasia, and liver/brain/pulmonary arteriovenous malformation).[6]

While JPS is inherited in an autosomal dominant manner in approximately 50–75% of cases, it can also arise sporadically in families without previous history of the disease.[7,8] The diagnosis of JPS is based on one of the following criteria:[4,8]

1. Five or more juvenile polyps in the colon.

2. Multiple juvenile polyps in the whole gastrointestinal tract (more than two organs).

3. Any number of juvenile polyps and family history of juvenile polyposis.

In our case, the patient met the diagnostic criteria for JPS by presenting with multiple juvenile polyps throughout her colon.

The majority of polyps in JPS are localized in the colon (particularly the rectosigmoid region), though they may also affect the stomach and, more rarely, the duodenum, jejunum, and ileum.[2] The number of polyps varies widely, from a few to as many as 50 to 200 in some families.[2] The term "juvenile" in JPS refers to the histological type of the polyp, which is characterized by abundant stromal tissue, cystic dilatation of the glands, and inflammatory cell infiltration.[7] Juvenile polyps develop from infancy through adulthood. While the age of onset varies, symptoms often emerge in the first or second decade of life, often as diarrhoea, rectal bleeding, prolapsing polyps, abdominal pain, intestinal obstruction (from intussusception), anaemia, anasarca, hypoproteinemia, protein-losing enteropathy, and failure to thrive.[2,4,9] Our patient first noticed rectal bleeding at the age of 5 and presented to our facility 11 years later with obvious growth retardation. Her failure to thrive and delayed puberty were likely consequences of chronic blood loss, anaemia, and protein-losing enteropathy due to her extensive polyp burden.

Juvenile polyposis syndrome can also present with extra-intestinal manifestations, spanning cutaneous manifestations (i.e., telangiectasia, pigmented nevi) and skeletal stigmata (i.e., polydactyly, hydrocephalus, hypertelorism and cleft palate) as the most frequently encountered (up to 56% and up to 70%, respectively).[6] These manifestations were not observed in our patient.

Multidisciplinary care, involving gastroenterologists, geneticists, and surgeons, is often required to manage the diverse manifestations of JPS, especially given the high cancer risk associated with this condition. Patients with JPS have a

cumulative colorectal cancer (CRC) risk of 39% to 68%, with a mean age of CRC developing estimated at 43.9 years in the John Hopkins polyposis registry data and 44 years in a large European retrospective study.[6] Approximately 21% of patients also face an increased risk for upper gastrointestinal cancers, including gastric, duodenal, and pancreatic cancer. Due to these risks, surveillance strategies are justified.

Guidelines from the European Society of Gastrointestinal Endoscopy (ESGE), American College of Gastroenterology (ACG), and European Society for Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN) recommend annual colonoscopy starting at age 12 years for colorectal polyps, with polypectomy for polyps larger than 5 mm, and upper endoscopy every 1–3 years beginning at age 18 years.[6,10,11] While the exact frequency of small bowel polyps is unknown, monitoring the small bowel is considered appropriate for symptomatic and anaemic JPS patients.

The primary treatment for JPS patients with a limited burden of polyps in any area of the GI tract is endoscopic polypectomy. In cases where polyps are numerous and difficult to control endoscopically, colectomy is considered, particularly when symptoms such as bleeding and diarrhoea are severe and there is suspicion of malignancy. [6,10,11]

With regards to the possible surgical treatments, colectomy with Ileo-rectal anastomosis or restorative proctocolectomy can be indicated. The choice of reconstructing an Ileal pouch is usually guided by the extent of rectal polyps, as recurrence has been reported in the remaining rectum or in the Ileal pouch. Oncel et al.[12] reported that five of ten patients who initially underwent subtotal colectomy with ileorectal anastomosis or partial colectomy required subsequent surgery within nine years due to symptomatic polyps, in situ carcinoma, or both; the remaining patients required multiple endoscopic polypectomies. Similarly, Hussain and Church[13] described a patient who developed polyps in an anal pouch after restorative proctocolectomy, ultimately necessitating pouch excision and end ileostomy. This patient also noted to develop polyps in the ileostomy during continued surveillance. These recurrences highlight the need for continued endoscopic follow up and monitoring of all operated patients, both in cases of restorative proctocolectomy and subtotal colectomy.[6,12] In our patient's case, we elected to perform a total proctocolectomy due to the presence of polyps in the rectum. The procedure was performed in stages to allow time for nutritional optimization and for the ileum to undergo adaptive changes before completing the final anastomosis. She was also counselled on the need for lifelong surveillance.

For JPS patients with extensive gastric polyposis, a complete or partial gastrectomy may be recommended when polyps are uncontrolled by endoscopic means or when gastric dysplasia is detected. Unlike Familial Adenomatous Polyposis, where NSAIDs such as sulindac or celecoxib can reduce colonic adenomas, no effective chemopreventive agent has yet been identified for JPS.[8] In addition, individuals with JPS and their families require genetic counselling and surveillance to understand the inheritance pattern and risks for future offspring. Family members may also undergo genetic testing to

determine whether they carry the same mutations and, if so, initiate early surveillance.

Juvenile Polyposis Syndrome (JPS) is a complex condition to manage, and the challenges are even more pronounced in resource-limited settings like ours. Our experience with this case highlights several obstacles we faced in diagnosing and treating this patient.

First, low awareness of rare conditions like JPS among both the public and healthcare workers was a barrier to early detection and intervention. This patient had visited multiple healthcare facilities over the years for her rectal bleeding, yet no diagnosis was ever made. Instead, she received repeated blood transfusions as a temporary measure to manage her anaemia.

Second, limited access to diagnostic tools hindered our ability to fully evaluate the patient. In well-resourced settings, total colonoscopy and upper GI endoscopy are the gold standards for diagnosis and surveillance of JPS. Without these tools, we could not assess the full polyp burden preoperatively, requiring us to make intraoperative decisions on bowel resection based on palpation and visual findings alone.

Third, genetic testing, which is essential for confirming diagnosis, assessing prognosis, and guiding family screening, was unavailable. Without testing for BMPR1A and SMAD4 mutations, we were unable to confirm the specific genetic variant or provide the family with necessary counseling and screening.

Additionally, financial constraints have further limited this patient's access to care. Her family relied on out-of-pocket payments, which made it difficult to afford the recommended treatments and follow-up care. As a result, the patient has been unable to keep up with the recommended surveillance schedule, and her family members have not come forward for examination or screening. This financial barrier is not unique to this case but is representative of a broader challenge in low-resource settings, where even basic healthcare services can be out of reach for many families.

Finally, ensuring essential lifelong follow-up is challenging in our setting, where limited healthcare infrastructure, scarce resources, and patient-related factors make consistent monitoring difficult.

In conclusion, Juvenile polyposis syndrome (JPS) is a rare but complex genetic condition with serious implications for gastrointestinal health and cancer risk. Advances in genetic testing have improved the ability to diagnose JPS and identify at-risk individuals; however, managing JPS remains challenging, requiring lifelong surveillance and, in some cases, surgical intervention. This case highlights the difficulties of managing JPS in a resource-limited environment, where the lack of advanced diagnostic tools and genetic testing often necessitates reliance on invasive procedures for diagnosis. Enabling access to such diagnostic resources would aid earlier detection, less invasive management, and more effective long-term surveillance. Addressing these gaps is important to improve outcomes for patients with hereditary polyposis syndromes in resource-constrained settings.

Consent: Informed consent was obtained from the patient and her mother to publish

this case report and the accompanying images.

Conflicts of interest: None.

REFERENCES

1. Wang LC, Lee HC, Yeung CY, Chan WT, Jiang CB. Gastrointestinal Polyps in Children. *Pediatr Neonatol.* 2009;50(5):196–201
2. Cauchin E, Touchefeu Y, Matysiak-Budnik T. Hamartomatous Tumors in the Gastrointestinal Tract. *Gastrointestinal Tumors.* 2015;2(2):65–74.
3. Gorji L, Albrecht P. Hamartomatous Polyps: Diagnosis, Surveillance, and Management. *World J Gastroenterol.* 2023; 29(8): 1304-1314
4. Haidle JL, MacFarland SP, Howe JR. Juvenile Polyposis Syndrome. *GeneReviews® - NCBI Bookshelf.* Published February 3, 2022.
5. Sayed MG, Ahmed AF, Ringold JR, Anderson ME, Bair JL, Mitros FA, et al. Germline SMAD4 or BMPRIA mutations and phenotype of juvenile polyposis. *Ann Surg Oncol.* 2002;9(9):901–906.
6. Dal Buono A, Gaiani F, Poliani L, Laghi L. Juvenile polyposis syndrome: An overview. *Best Pract Res Clin Gastroenterol.* 2022;58-59:101799.
7. Cao K, Plazzer JP, Macrae F. SMAD4 variants and its genotype–phenotype correlations to juvenile polyposis syndrome. *Hereditary Cancer in Clinical Practice.* 2023;21(1).
8. Matsumoto T, Umeno J, Jimbo K, Arai M, Iwama I, Kashida H, et al. Clinical Guidelines for Diagnosis and Management of Juvenile Polyposis Syndrome in Children and Adults-Secondary Publication. *J Anus Rectum Colon.* 2023;7(2):115–125.
9. Taylor H, Yerlioglu D, Phen C, Ballauff A, Nedelkopoulou N, Spier I, et al. mTOR inhibitors reduce enteropathy, intestinal bleeding and colectomy rate in patients with juvenile polyposis of infancy with PTEN-BMPR1A deletion. *Hum Mol Genet.* 2021;30(14):1273–1282.
10. Syngal S, Brand RE, Church JM, Giardiello, FM, Hampel HL, Burt RW. ACG Clinical Guideline: Genetic Testing and Management of Hereditary Gastrointestinal Cancer Syndromes. *Am J Gastroenterol.* 2015;110(2):223–262.
11. Cohen S, Hyer W, Mas E, et al. Management of Juvenile Polyposis Syndrome in Children and Adolescents. *J Pediatr Gastroenterol Nutr.* 2019;68(3):453–462.
12. Oncel M, Church JM, Remzi FH, Fazio VW. Colonic Surgery in Patients With Juvenile Polyposis Syndrome: A Case Series. *Dis Colon Rectum.* 2005;48(1):49–56.
13. Hussain T, Church JM. Juvenile polyposis syndrome. *Clin Case Rep.* 2019;8(1):92–95.