JUVENILE POLYPOSIS SYNDROME PRESENTING AS INTUSSUSCEPTION IN A MALNOURISHED CHILD: A CASE REPORT

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ABSTRACT
We report a rare case of juvenile polyposis syndrome (JPS) in a 5 year old girl who was admitted into the emergency room on account of acute intestinal obstruction secondary to ileo-colic intussusception. Prior to the admission, she had had repeated blood transfusion in the referring hospital following anaemia due to long standing rectal bleeding with malnutrition. The obstruction was relieved following ileocolectomy. Examination of the sample showed innumerable carpet-like polyps and 18 pedunculated polyps in the ascending and transverse colon. All the polyps have microscopic features consistent with juvenile polyps with no evidence of atypia. She was discharged a week after surgery and follow-up at outpatient clinic showed sustained clinical improvement. We discuss management challenges and follow-up plan in this patient with juvenile polyposis syndrome in view of its associated risk of gastrointestinal carcinoma as well as screening of the other family members for mutations linked to JPS.

Keywords: Juvenile polyposis, Intussusception, Anaemia, Colon, Mutation

INTRODUCTION
Juvenile Polyposis Syndrome (JPS) is a rare disease characterized by the presence of 5 or more juvenile polyps in the colorectum, juvenile polyps throughout the gastrointestinal tract or any number of juvenile polyps with a positive family history.1,2

Juvenile polyposis syndrome is one of the four hamartomatous-polyposis syndromes that include Peutz-Jeghers, Cowden, and Bannayan-Riley-Ruvalcaba syndromes.3 JPS is inherited in an autosomal dominant pattern occurring in 1:100 000 and entails an increased risk of colorectal carcinoma and to a lesser degree gastric cancer.4

JPS is associated with germline mutations in one of two genes connected with TGFβ/ BMP signal pathway.2,5 These mutations include point mutations and small deletions/insertions that were initially found in the tumor suppressor gene SMAD4 in several families. Other mutations were identified in the bone morphogenetic protein receptor type 1A (BMPRIA), a receptor involved in the BMP signaling pathway, of which SMAD4 is the intracellular mediator.5
Consequences of these mutations include loss of regulation of colonic mucosal development leading to unhindered mucosal growth and polyps in the gastrointestinal tract.

The index patient presented with acute intestinal obstruction from florid colonic mucosal polyposis that had caused chronic rectal bleeding and protein losing gastroenteropathy.

**CASE REPORT**

The patient was Miss M.N, a 5 year old girl who presented with a history of recurrent bloody stool for 3 years, weight loss and abdominal distention for 1 year. She was initially seen at a general hospital with a history of progressive body weakness and was found to be anaemic which necessitated her admission and blood transfusion. The patient however defaulted, only to present at our Paediatrics emergency 8 weeks after referral when she developed abdominal pains accompanied by vomiting. On admission, she was underweight (15kg), pale, febrile but anicteric. There was grade 4 finger clubbing. The abdomen was distended with obvious bowel markings and a mass in the right lumbar region measuring 6x5cm. The bowel sound was hyperactive. The respiratory, cardiovascular and central nervous systems were unremarkable. An initial clinical diagnosis of lymphoproliferative disorder to exclude intraabdominal malignancy was made.

Admission full blood count was normal and together with the blood film appearances, a history of progressive body weakness and was found to be anaemic which necessitated her admission and blood transfusion. She was thereafter referred to our health facility being a tertiary hospital after a repeat lymphoproliferative disease was unlikely. HIV I and II were negative. Abdominal ultrasound was reported as ileo-colic intussusception and this was confirmed by CT scan (Figure 1a&b).

Fig. 1a: CT scan of the abdomen showing invagination of the ileocolic segment into ascending colon giving a complex heterogenous mass (arrow) in the region of hepatic flexure. **Fig 1b:** CT scan showing dilated bowel loops with air fluid levels.
She had exploratory laparotomy and resection of the intussusception that involved the distal 30 cm terminal ileum, caecum, ascending colon and proximal 2/3 of transverse colon. An end-to-end anastomosis was achieved. She had bilateral pedal swelling up to the thigh noted on the 3rd day post-operative (DPO).

The post-operative PCV and repeat E, U & Cr were within normal range but total protein was low. On the 5th DPO, she was commenced on high protein diet and by 8th DPO, the pedal swelling had resolved and she was discharged to the outpatient clinic. Four weeks after being discharged from the ward, her weight increased to 20 kg (25% increase over admission weight) and there was also clinical improvement.

**Histopathological findings**

**Macroscopy:** The specimen (Fig. 2a) consisted of terminal ileum, appendix, caecum, ascending colon and proximal part of transverse colon. The appendix together with parts of terminal ileum and caecum telescoped into the ascending colon. The cut section showed hyperaemic mucosal lining carpeted by small sized polyps and 18 pedunculated polyps (sizes ranged from 1.0-1.5 cm) covering the entire colonic mucosal lining from the ileocaecal junction to the distal resection margin.

**Microscopy:** The colonic polyps are similar showing variably sized glands that are lined by columnar epithelial cells surrounded by inflamed, oedematous lamina propria (Fig 2b). The diagnosis of juvenile polyposis syndrome was made.

**Follow-up and management:** The patient had sustained clinical improvement 6 months post-surgery. The parents were informed on the need to know the patient and her siblings' mutation status through screening for SMAD4 or BMPR1A. The need for lower and upper gastrointestinal endoscopy in view of the presence of polyps at the distal resection margins was also emphasized.

**DISCUSSION**

Juvenile polyposis syndrome is an autosomal dominant disorder characterized by the development of multiple hamartomatous polyps in the gastrointestinal tracts and 20-50% of affected patients have a positive family history. The case presented has numerous polyps in the colon but no positive family history of juvenile polyposis. Mutational analysis in patients with juvenile polyposis detects mutations in BMPR1A in 20-30% of patients, and in SMAD4 in

![Fig. 2a: Ileo-colic bowel segment showing numerous small-sized sessile and pedunculated polyps covering the entire colonic mucosal lining.](image1)
![Fig. 2b: Microscopic section of the colonic polyp shows variably sized glands some of which are dilated surrounded by oedematous inflamed stroma. Haematoxylin & Eosin x4.](image2)
20-30% of patients. Our patient is yet to have mutation screening done due to cost implication since it has to be carried out in genetic testing laboratory outside Nigeria.

Generally, colorectal polyps present commonly as rectal bleeding in children with or without abdominal pains. Our patient had a long history of rectal bleeding with resultant anaemia which necessitated repeated blood transfusion. This is similar to the observation in 1 out of the 16 cases of juvenile polyps reported by Anyanwu et al. in Kano. Removal of such polyps via colonoscopy is a means of preventing intermittent polyp bleeding, prevent anaemia and reduce the potential risk of malignant transformation subsequently.

Intussusception may also develop from gastrointestinal polyps if the polyp extrudes sufficiently into the lumen, propelled by peristalsis and traction. This could lead to acute intestinal obstruction and presentation of abdominal pains as was seen in the presented case. In many documented cases of intussusception, management is dependent on the cause, and interval between onset of symptoms and presentation in the hospital. Surgical treatment with enterostomy and polypectomy or segmental bowel resection to remove the lead point of the intussusception is required. The case presented had segmental resection of part of terminal ileum, caecum and ascending colon with ileocolic end to end anastomosis.

JPS is associated with an increased lifetime risk for both colorectal cancer (CRC) and gastric cancer. Bronsen et al. in a recent cancer risk analysis calculated a cumulative life-time risk for colorectal cancer in JPS of 39% and a relative risk of colorectal cancer of 34. To reduce this risk, patients with JPS are expected to have surveillance endoscopy every 2 – 3 years or yearly in those with polyps until they are deemed polyps-free. If the polyps are fewer, these are removed via endoscopy while in those with many polyps or severe symptoms, partial colectomy or total colectomy is offered depending on the extent of the disease.

Genetic analysis to test for the presence of mutations in SMAD4/ BMPR1A would have further supported the clinical findings, confirm the diagnosis and probably evaluate the risk for colorectal cancer in this patient. Due to unavailability and cost, we were unable to carry out genetic analysis in this patient.

REFERENCES