An Unusual Presentation of Peutz-Jeghers Syndrome: Recurrent Intussusception in a 13-Year-Old Female: A Case Report

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INTRODUCTION

Peutz Jeghers syndrome an autosomal dominant condition due to mutation in chromosome 19p13.3 can present with intestinal hamartomatous polyps. Typically, they present with hyperpigmented macules in the mucosa. We present a case of a 13-year-old girl who presented in emergency department with recurrent right sided abdominal pain and tenderness, vomiting, and a palpable mass. Patient has a significant positive family history of hyperpigmented macules. Patient was evaluated with contrast enhanced CT scan and was found to have ileo-ileal intussusception and a gastric fundic polyp. Patient underwent emergency laparotomy and was found to have Jejunojejunal intussusception with polyp at lead point.

PRESENTATION OF CASE

A 13-year-old girl presented in emergency department with complaints of abdominal pain, nausea & vomiting and constipation of one day duration. The patient was having recurrent episodes of abdominal pain over the past 2 month which previously got relieved after vomiting. There was also history of two episodes of blood in stools over the past 2 months. There was no history of loss of weight or appetite and no alteration in bowel habits.

On general examination there were multiple well demarcated hyper pigmented macules on the lips tongue and face. Her father and sister also had similar lesions on the face and lips. Rest of the skin, hair nails trunk extremities gums & palate was normal.

Figure 1.
Oral Lesion

On examination abdomen was distended with diffuse tenderness but no rebound tenderness. There was a well-defined tender mass in right iliac and lumbar region.

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CLINICAL DIAGNOSIS

Acute Intestinal Obstruction: Intussusception

DIFFERENTIAL DIAGNOSIS

- Intussusception due to Intestinal Polyps in Peutz Jeghers Syndrome.
- Intussusception due to Meckel's Diverticulum.

INVESTIGATIONS AND MANAGEMENT

Ultrasonography showed bowel in bowel appearance consistent with intussusception. CT abdomen showed small bowel intussusception, possibly ileo-ileal along with a gastric fundic polyp.

Figure 2. CECT Showing Intussusception (Cursor)

An emergency laparotomy showed a jejuno-jejunal intussusception with a pedunculated polyp at 65 cm from the DJ flexure. There was also another non obstructive polypoidal lesion in the proximal transverse colon which is planned to be snared after 2 months through colonoscopy.

Intussusception has a peak incidence in the age group of 5 months - 10 yrs. And is mostly idiopathic in infancy and after 2 years of age 1/3rd of the cases have a lead point[1] Adolescent intussusceptions are due to mainly by Meckel's diverticular inversion and very rarely polyps as a lead point. Peutz Jeghers syndrome an autosomal dominant condition due to mutation in chromosome 19p13.3 can present with intestinal hamartomatous polyps. Two third of these patients experience an intussusception during their lifetime, mostly in small bowel.[2]

Here we are presenting a case of 13-year-old girl who had recurrent attacks of abdominal pain and diagnosed to have intussusception secondary to Peutz Jeghers syndrome which was suspected in view of mucosal pigmentation. This highlights the importance of the awareness of this condition in adolescent intussusceptions.

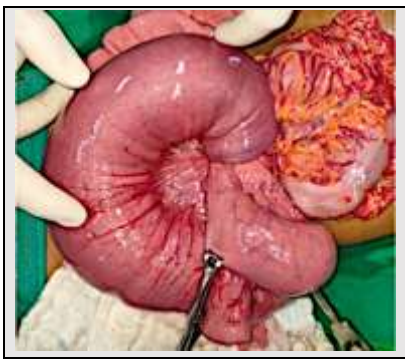

Figure 3. Intussusception - Intraoperative Image Showing Polyp as the Lead Point

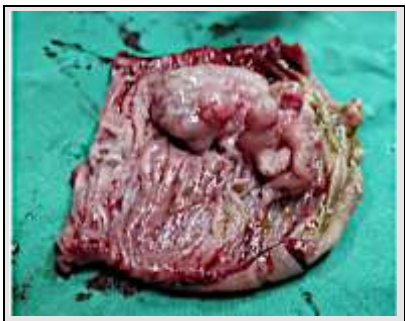

Figure 4. Gross Specimen

PATHOLOGICAL DISCUSSION

Peutz-Jeghers syndrome (PJS) is an autosomal dominant syndrome with 905 penetrance. It is due to germline mutations in the STK11 (LKB1) gene encoding a serine threonine kinase mapped to chromosome 19p13.3 characterized by multiple hamartomatous polyps in the gastrointestinal tract, mucocutaneous pigmentation, and an increased risk of gastrointestinal and non-gastrointestinal cancer. It is a rare condition with an estimated prevalence of 1: 8000 to 1: 200,000 births with no sex predilection.[3] Our patient meet all the three clinical criteria for PJS viz. Two Peutz-Jeghers-type hamartomatous polyps of the gastrointestinal tract, Mucocutaneous hyperpigmentation of the mouth, lips, and Family history of PJS.

PJS typically is characterised by pigmented mucocutaneous freckles of lips and multiple hamartomatous gastrointestinal polyps. Individuals with PJS are also at an increased risk for both gastrointestinal and extra-intestinal cancers. Although polyps most commonly occur in the small bowel (60 to 90 percent) and more specifically in the jejunum, they can be found throughout the gastrointestinal tract including the stomach (15 to 30 percent) and colon (50 to 64 percent).[4]

Over half of the patients are asymptomatic at the time of diagnosis. Individuals with PJS can present with obstruction caused by intussusception or occlusion of the gastrointestinal lumen by the polyp, abdominal pain caused by infarction, anaemia from acute or chronic bleeding, or extrusion of the polyp through the rectum.

Intussusception can occur in almost two third during their lifetime, most often is Jejunojejunal Intussusception. PJS is also associated with an increased risk of gastrointestinal and extra-intestinal malignancies. In a systematic review of 20 observational studies which included

1644 patients with PJS, lifetime risk for any cancer varied between 37 and 93 percent. The risk of extra-intestinal cancers is also increased in individuals with PJS breast, ovary and cervix in women and Sertoli cell testicular tumours in men.

Intussusception is frequent in children of 5-10-years age group. However, bowel intussusception in older children & adults is considered a rare condition, accounting for only almost 1%-5% of bowel obstruction.[5]

Adolescent intussusceptions will have a lead point and in the small intestine, an intussusception can be secondary either to the presence of intra- or extra-luminal lesions like inflammatory lesions, Meckel's diverticulum, postoperative adhesions, lipoma, adenomatous polyps, lymphoma and metastases.[6] Peutz Jeghers polyps are rarely considered as a primary cause unless the general findings are attributable.[7]

FINAL DIAGNOSIS

The presence of mucocutaneous macules with freckling and a strong family history are the key factors in deciding the further course in even asymptomatic patients with PJS and the final diagnosis of acute intestinal obstruction with jejunal polyps in Peutz Jeghers Syndrome. Intussusception in adults and older children is rare contributing to only 5% of the intussusceptions and hamartomata's polyps seen in Peutz Jeghers syndrome can act as lead points and cause intussusception.

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