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A Rare Case of Peutz Jeghers Syndrome Presented with Recurrent Multiple Intussception

Dr. Shivam Maheshwarl[1], Dr. Arjun Pawar[2]

[1]JR2, MGM Medical College, Chh. Sambhajinagar

[2]A Professor, Pediatric Surgeon, MGM Medical College, Chh. Sambhajinagar

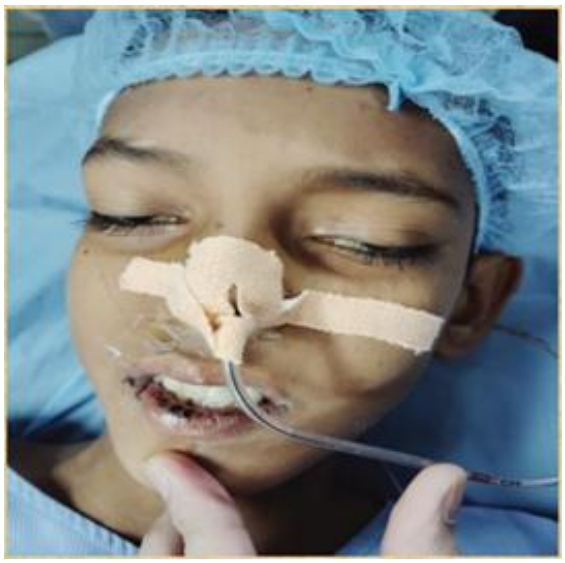
Abstract: ***Peutz-Jeghers syndrome is an uncommon inherited condition marked by gastrointestinal polyps and characteristic mucocutaneous pigmentation, often linked to mutations in the STK11 gene. A case involving an eleven-year-old boy is presented, with a prolonged history of intermittent colicky abdominal pain and weight loss, previously managed conservatively for recurrent small bowel intussusception. On current admission, the patient showed persistent symptoms along with vomiting and visible pigmentation over the lips and buccal mucosa. Imaging revealed multiple sites of small bowel intussusception with signs of compromised blood supply. Surgical exploration identified jejunal and jejunoileal intussusceptions, both reduced manually, with a large polyp serving as the lead point. Resection and anastomosis were performed, and additional polyps detected during intraoperative assessment were excised. Histopathological evaluation confirmed the diagnosis of Peutz-Jeghers syndrome. The postoperative course remained stable, and further endoscopic evaluation was planned to assess additional gastrointestinal involvement.***

Keywords: Peutz Jeghers syndrome, intussusception, gastrointestinal polyps, mucocutaneous pigmentation, pediatric bowel obstruction

1. Introduction

Peutz-Jeghers syndrome is a rare autosomal dominant genetic disorder characterized by:

- **Hamartomatous gastrointestinal polyps**
- **Mucocutaneous pigmentation**
- Caused by mutation in the STK11 (LKB1) gene



Intussuscetion is a condition in which **one segment of intestine telescopes into an adjacent segment,** leading to bowel oostruction and impaired blood flow.

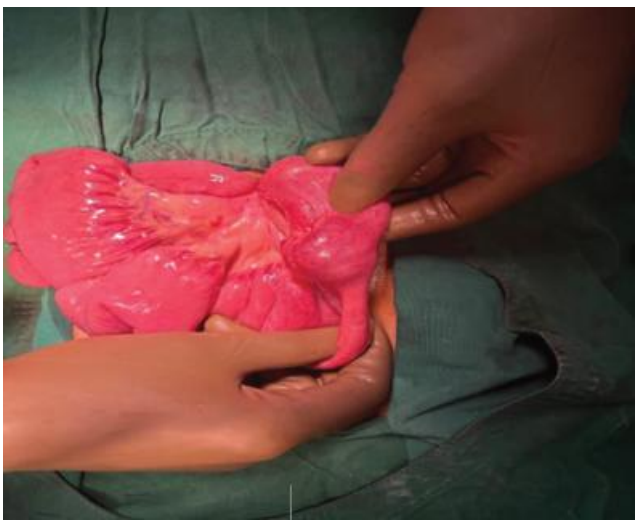
4°/o cases have some pathological lead point like polyp, Meckels, lymphoma, duplications cyst etc.

Rare in adolescent- if occurs-usually has got some pathological lead point.

2. Case Report

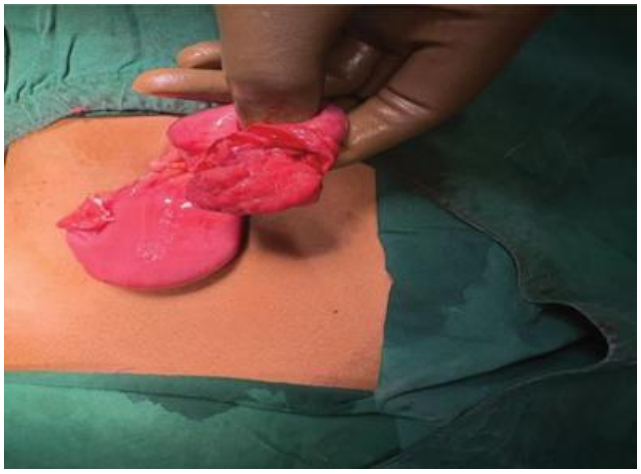
- A 11- year old boy presented with Recurrent abdominal pain, colicky in nature for 2 years and weight loss at various private hospital. On evaluation diagnosed to have small bowel intussusception and was managed conservatively on multiple occasions.
- At our institute patient presented with similar complaints along with multiple episodes of vomiting.
- On examination purple pigmentation was observed over lips and buccal mucosa.
- PlA- Palpable lump in left side of abdomen. No tenderness USG finding s/o intussusception
- CECT abdomen- showed small bowel intussusception at two sites with compromised vascularity.

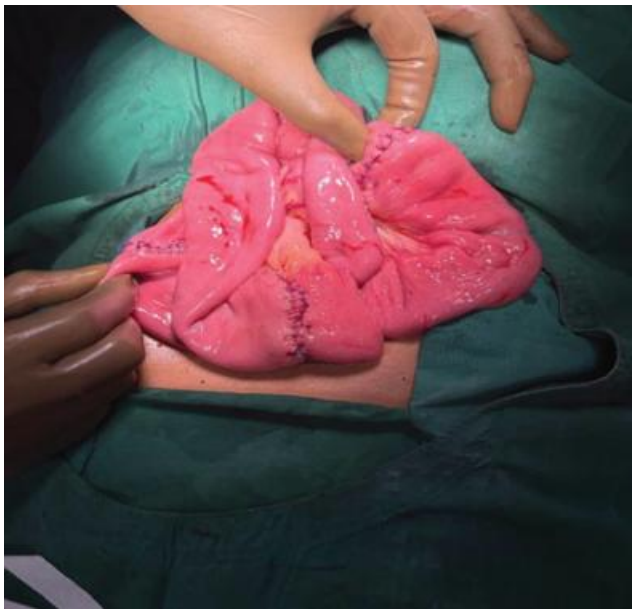
3. Operative Details



- Vertical midline laparotomy was performed.
- E/O Two long segment intussusception- jejunal and jejunoileal intussception was found.
- manual reduction done.
- Bowel was viable.
- Large polypoidal growth was acting as PLP.
- Resection anastomosis was performed.
- Entire bowel palpated for polyp at any other site. One

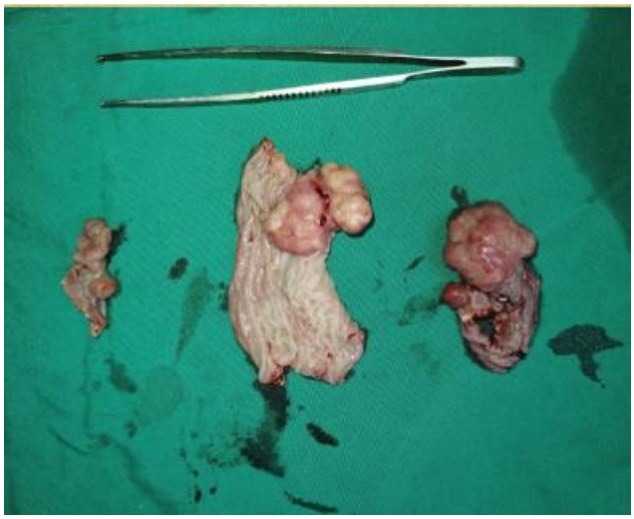
more 1 cm polyp was found and removed by wedge resection.





Three palpable polyp

- at 15 cm from DJ flexure
- 90 cm from DJ flexure
- 120 cm from DJ flexure
- 3 Polyps were sent for Histopath examination



- Later the Histopath report came as- Peutz Jeghers syndrome.
- Follow up- Asymptomatic.
- Planned for Upper GI scopy and colonoscopy.

4. Summary

Peutz-Jeghers syndrome is an uncommon inherited condition marked by gastrointestinal polyps and characteristic mucocutaneous pigmentation, often linked to mutations in the STK11 gene. A case involving an eleven year old boy is presented, with a prolonged history of intermittent colicky abdominal pain and weight loss, previously managed conservatively for recurrent small bowel intussusception. On current admission, the patient showed persistent symptoms along with vomiting and visible pigmentation over the lips and buccal mucosa. Imaging revealed multiple sites of small bowel intussusception with signs of compromised blood supply. Surgical exploration identified jejunal and jejunoileal intussusceptions, both reduced manually, with a large polyp serving as the lead point. Resection and anastomosis were performed, and additional polyps detected during intraoperative assessment were excised. Histopathological evaluation confirmed the diagnosis of Peutz-Jeghers syndrome. The postoperative course remained stable, and further endoscopic evaluation was planned to assess additional gastrointestinal involvement.

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