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A CASE REPORT OF PEUTZ -JEGHERS SYNDROME PRESENTING AS MULTIPLE INTUSSUSCEPION

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Abstract : Peutz-Jeghers syndrome is a rare familial autosomal dominant disorder. Peutz -Jeghers syndrome in a young girl presented as multiple intussusceptions involving small bowel. Complaints were intermittent abdominal pain with minimal abdominal signs. She was managed with laparotomy, and the findings were three intussusceptions involving the ileum. Peutz- Jeghers syndrome itself rare which presented as three intussusceptions with subtle signs of obstruction is rare.

Keyword : Peutz - Jeghers syndrome, Intussusception, hamartamatous polyp, entrostomy, laparotomy

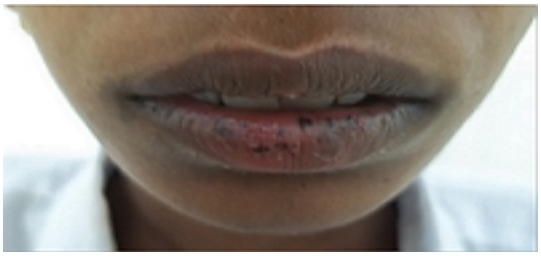
Introduction

Peutz- Jeghers syndrome is a rare familial autosomal dominant disorder characterised by mucocutaneous pigmentation, gastrointestinal and extragastrointestinal hamartamatous polyps and increased risk of malignancy. Incidence is 1 in 60,000 to 3,00,000 live births.[1][2] Germ line mutation involving the STK 11/LKB1 tumour suppressor gene located in band 19p 13.3 .[3][4] Of the many published case reports of Puetz-Jeghers syndrome many patients presented with bleeding and intussusceptions. We present the case for the rarity in the general population, subtle presentation of intestinal obstruction, and multiple intussusceptions intraoperatively.

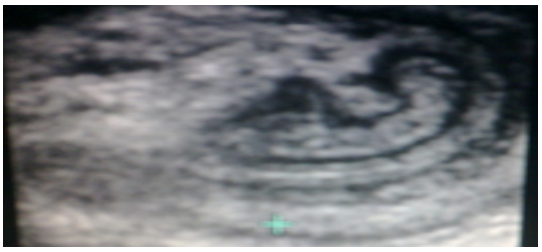
Case presentation

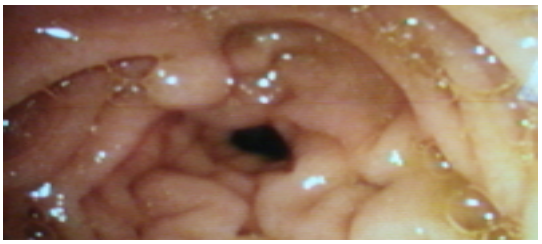
A 14 year young girl presented to our emergency department with history of abdominal pain for the past two days. Pain was colicky in nature, without radiation and not associated with vomiting .But not passed flatus and motion for 1 day. No history of melena .No previous history of similar illness in the past. There was no any previous surgery in the past.

She was very thin built, her vitals were stable, abdomen was soft and scaphoid, without a mass and localised tenderness. Bowel sounds were present. Rectal examination was normal. Multiple dark pigmented patches were seen on the buccal mucosa and in the perianal region.

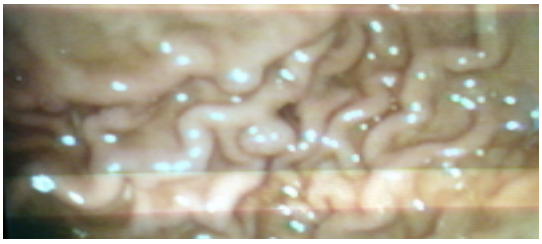


MUCOCUTANEOUS PIGMENTATION





ULTRASOUND WHORL SIGN



COLONOSCOPY SHOWING POLYP

U G I S C O P Y S H O W I N G P O L Y P

X ray doesn't show air fluid levels or dilated bowel loops. Ultrasound abdomen was taken which showed the whorl sign. Computerised tomography of abdomen was repoted as small bowel obstruction. Upper gastrointestinal scopy was done which showed multiple polyps in the gastric mucosa which are nearly 0.5 to 1 cm in size. Colonoscopy was done which showed multiple pedunculated polyps in the large bowel.

Management and outcome

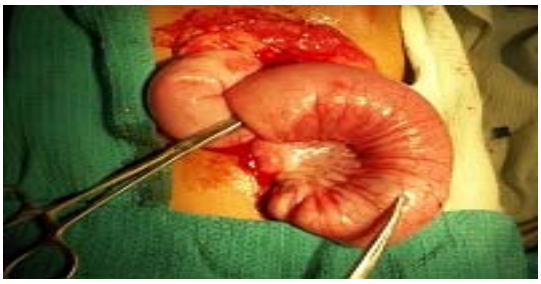
Initially planned for conservative line of management. Since even after 1 week of conservative management there was no relief of symptoms and planned for a laparotomy and proceed.

She had three intussusceptions in the small intestine which are ileoileal with no dilatation of nproximal bowel segments. Two of the three intussusceptions were reduced and the lead points (polyp) were removed by entrostomy. The polyps were 1 x 1 cm in size. But the third intussusception could not be reduced and bowel wall was unhealthy, so resection and anastomosis of that segment done. The resected segment had a polyp of size 4 x 3 cm . Other segment of small bowel was traced for palpable polyps but none was big enough to be palpable. Postoperative period was uneventful.

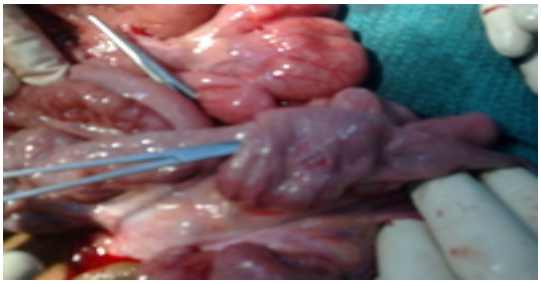
Biopsy was reported as hemartamatous polyp. Colonoscopy and upper gastrointestinal scopy was done for their parents and siblings. None had polyps in the gastro intestinal tract. Patient came for follow up for nearly 1 year with history of occasional colicky pain which relieved by itself.

In this case we have removed only three large polyps in the small intestine which acted as lead points for intussusception. Two of the polyps were removed by seperate enterotomy .The third intussusception could not be reduced and the bowel wall was found to be unhealthy so resection and anastomosis was done.

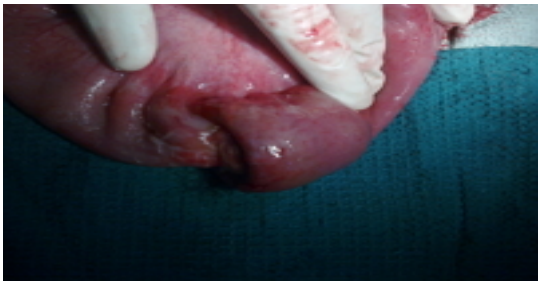
Polyps in peutz jegher syndrome which are asymptomatic and less than 1.5 cm can be left untouched as these patients will have numerous polyps through out the gastointestinal tract and all cant be removed.Only symptomatic polys or polyps greater than 1.5 cm has to be removed during laparotomy. During colonoscopy or upper gastrointestinal scopy polyps greater than 0.5 cm has to be removed



INTUSSESCEPTION 1 .

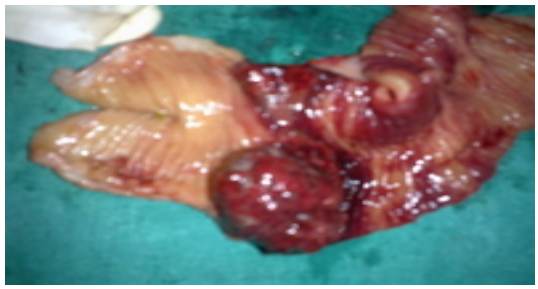


INTUSSUSCEPTION 2

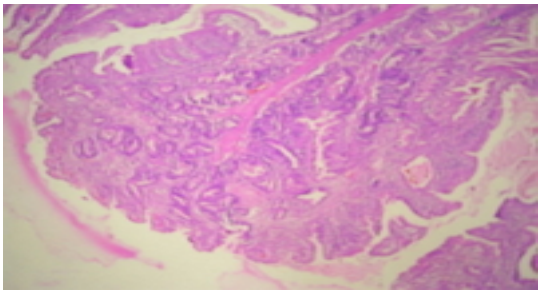


INTUSSUSCEPTION 3

DISCUSSION Intussusception is telescoping of one segment of gastrointestinal tract in to another. Enteric intussusceptions in adults are rare with a pathological lead point in 90% of the cases. Ileoileal intussusceptions is the most common type.[5] About 90% of cases presents with abdominal pain ,40% presents with obstruction. Rare presentations were melena, bleeding per rectum, diarrhoea,



CUT SECTION SHOWING POLYP



BIOPSY-HAMARTAMATOUS POLYP

Peutz- Jegher syndrome is an autosomal dominant inherited syndrome characterised by intestinal hemartamatous polyp and mucocutaneous pigmentations. Patients with Peutz -Jegher syndrome have 15 fold increased risk of developing intestinal cancer than general the population. ([7] [8][9]The mean age for first diagnosis of cancer was 43+/-10 years. [10][11][12]The median time for first time for polyp to occur is 10 to 13 years .The incidence of polyp is greatest in the jejunum and progressively decreases in the ileum and duodenum.[(2)]

Most frequent presentation is recurrent abdominal pain caused by obstruction and transient intussescepions. Melena and rectal bleeding occur less frequently .Most studies show that nearly 50% of patients experience intussusceptions during their lifetime. [(13 Mucocutaneous pigmentations and melanin spots are typically present in 95% of patients. Cutaneous pigmentations are locted in the perioral, perinasal, perianal region and genitals.World health organisation clinic- pathological criteria for diagnosing Peutz -Jeghers syndrome (14

1. Three or more polyps, which show pathological features consistent with Peutz- Jeghers syndrome.
2. A family history of Peutz -Jeghers syndrome with any number of Peutz-Jegher polyps.
3. A family history of Peutz -Jeghers syndrome with characteristic mucocutaneouspigmentation.
4. Mucocutaneous pigmentations with any number of Peutz -Jegher polyps.

Gastrointestinal polyps seen in Peutz -Jeghers syndrome are hemortamatous polyps. Histology is characterised by extensive smooth muscle arborisation throughout the polyp .It is associated with increased incidence of not only gastrointestinal adenocarci noma but also extraintestinal malignancyof the breast, pancreas, testes, ovary.(7) In most of the case reports Peutz-Jegher syndrome presented with melena or intussusceptions.

(15)But in this patient the presentation was with multiple intussesceptions with a completely unremarkable abdominal examination.

Conclusion

In patients with Peutz- Jegher syndrome even if the symptoms and clinical examination were normal we have to investigate further to identify subtle intestinal obstruction .During surgery we have to examine the whole intestine to rule out or identify other intussesception and palpable polyps which can be removed in same laparotomy so further laparotomies can be reduced as these patients usually undergo numerous surgeries in their life time.

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