

CASE REPORT

INTUSSUSCEPTION IN A CASE OF PEUTZ JEGHER SYNDROME: A CASE REPORT

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ABSTRACT: PeutzJegher syndrome is a rare disorder characterized by mucocutaneous melanin deposits over lips, oral mucosa, fingers and multiple hamartomatous polyps in gastrointestinal tract. These individuals may present with small intestinal intussusception with these polyp as lead point. We report a case of 15 year old boy presenting with intestinal obstruction with oral melanin deposits and found to have multiple hamartomatous polyps in small intestine.

KEYWORDS: Peutzjegher syndrome, intussusception, mucocutaneous pigmentation, hamartomatous polyps.

INTRODUCTION: Peutzjeghers syndrome is a rare autosomal dominant disorder with variable penetrance caused by a mutation in STK/LKB1 tumour suppressor gene. It is characterised by intestinal hamartomatous polyps in association with mucocutaneous melanocytic macules on the lips and oral mucosa.

We present a case of 15 year old male patient with mucocutaneous hyperpigmentation and small bowel obstruction due to jejunojejunal intussusception with peutzjeghers polyps treated by lap-assisted intestinal resection.

CASE REPORT: A 15 year old male individual presented with abdominal pain and 4 episodes of bilious vomiting for 2 days with history of intermittent attacks of abdominal pain and vomiting over past 1 month. No history of bleeding per rectum. Family history is not significant. On physical examination there was hyperpigmentation of lips and buccal mucosa along with signs of dehydration. On palpation of abdomen there was tenderness over left hypochondriac and para umbilical region. No guarding or rigidity. No palpable masses are noted and exaggerated bowel sounds are noted.

Blood investigations are unremarkable. X-ray abdomen in erect posture showed dilated bowel loop in left lumbar and umbilical region. Ultrasound abdomen showed pseudo-kidney sign in left para umbilical region suggestive of intususception. Lap assisted resection anastomosis was planned.

Intra op findings were dilated jejunal loop with jejuno-jejunalintususception is noted 1 feet from duodeno-jejunal flexure. Laparoscopic reduction followed by laporotomy with resection of involved segment andjejuno-jejunal anastomosis was done. Resected segment showed multiple jejunalpolyps and the specimen was sent for HPE. Post –op recovery is uneventful.

HPE report is suggestive of Peutzjeghers polyps with no evidence of malignancy. Hamartomatous polyp of jejunum showing dilated glands devoid of atypiaseparated by bundles of smooth muscle.

CASE REPORT

DISCUSSION: Peutz-Jeghers syndrome first described by Jan Peutz and Harold Jeghers independently as generalized polyposis and melanin spots of oral mucosa, lips and digits. It has incidence of 1 in 8300 to 1 in 28000 individuals,¹ however incidence varies with geographical location. It is inherited as autosomal dominant condition with variable penetrance. Mutation in STK/LKB1 gene located on 19p13.3 is responsible for 66-94% of cases.² Sporadic cases are also reported with increasing incidence. Mehnani et al reported an increasing mutational spectrum of allelic variants of STK/LKB1 gene worldwide responsible for increasing incidence of this condition.²

The median age of onset for GI symptoms is 13 years and by 20 years of age 50% of individuals will have anemia, rectal bleeding, abdominal pain, obstruction and/or intussusception.^{3,4} Nearly, 50% of patients experience intussusception during their life time.⁵

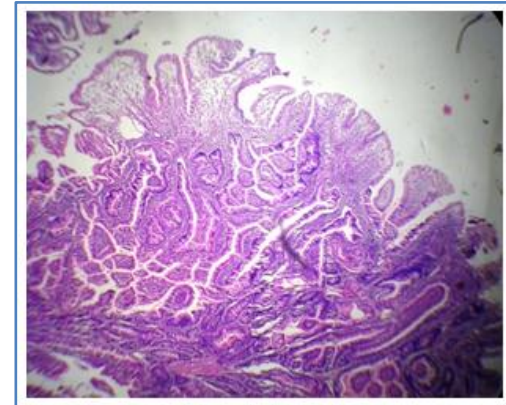
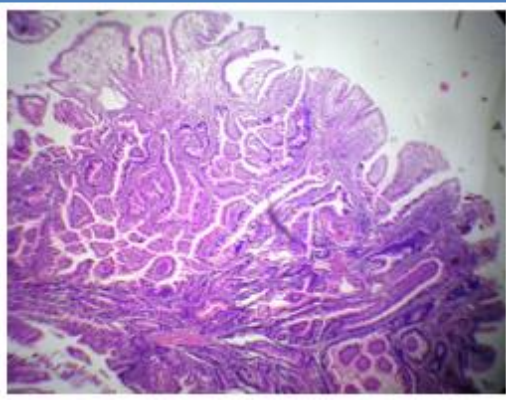
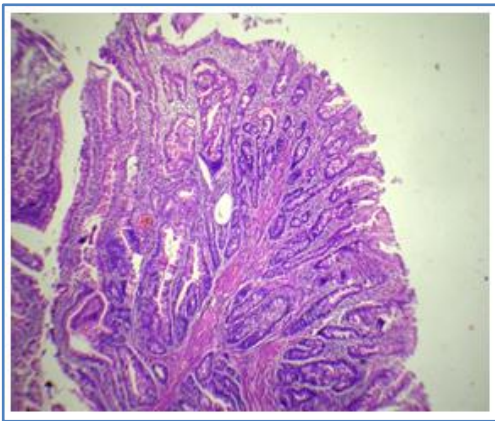
Multiple hamartomatous polyps are seen in gastro intestinal tract along with mucocutaneous pigmented lesions. Utsunomiya study showed polyps are seen with frequency in small intestine (64%), colon (63.2%), stomach (48.6%) and rectum (32%). In small intestine, polyps are more common in jejunum followed by duodenum and ileum.⁶⁻¹¹ Extra intestinal polyps are also reported in gall bladder,¹² nose,¹³ ureter.¹⁴ The most characteristic feature of a Peutz-Jeghers polyp is a central core of smooth muscle that extends into the polyp in an arborizing fashion (Christmas tree like appearance) and that is covered by either normal or hyperplastic mucosa native to the involved site. Polyps can be pedunculated, sessile varying in size from subcentimeter to more than 3.5 cm and these polyps can form a lead point for intussusception and cause obstruction. Obstruction can also be caused when polyps grow large.

The risk of malignant transformation of these polyps though less compared to other intestinal polyposis conditions, it is significantly high when compared to general population. The risk of malignancy increases with age. Both intestinal and extra intestinal malignancies like breast, cervical, testicular, lung.¹⁵ The overall relative risk for cancer is greater in females than in males and greatest for gastrointestinal, pancreatic, and gynecologic cervical cancers.¹⁶

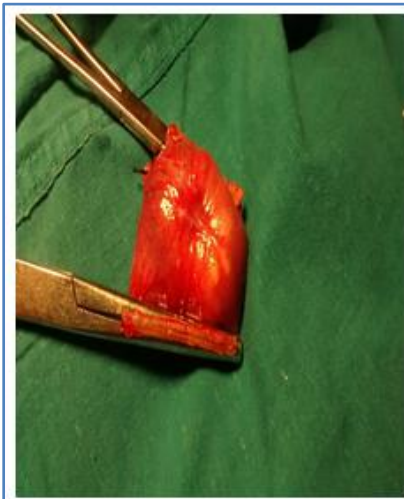
The diagnosis of Peutz-Jeghers syndrome can be made by presence of any two of these 3 features like multiple hamartomatous polyps in intestine, mucocutaneous pigmentation and significant family history. Imaging when done for gastro intestinal symptoms may demonstrate multiple polyps throughout the intestine. No specific laboratory investigations help in making the diagnosis but genetic screening in first degree relatives may help identify at early age.

Treatment of patients presenting with acute gastrointestinal symptoms is laparotomy and resection of involved intestine containing polyps responsible for symptoms. Endoscopic polypectomy of any hemorrhagic or large symptomatic polyp should be done after the diagnosis. Surgery or laparoscopy combined with intra operative enteroscopy is indicated for those identified with this condition at an early age and may require multiple operations and resection of intestine that can lead to short bowel syndrome.¹⁷ Routine breast and gynecologic surveillance along with routine periodic surveillance is needed to identify malignancies at an early age.

CASE REPORT



Separated by bundles of smooth muscle



CASE REPORT



Peutz jeghers polyps forming a lead point for jejuno-jejunal intussusception

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CASE REPORT

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