

PEUTZ JEGHERS SYNDROME PRESENTING WITH ACUTE INTESTINAL OBSTRUCTION: A RARE CASE REPORT WITH REVIEW OF LITERATURES

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Background: Peutz-Jeghers syndrome is a rare genetic disorder presenting in young age with mucocutaneous pigmentation and hamartomatous polyposis. **Method:** We report a case of Peutz-Jeghers syndrome in a 16 year old boy presenting with acute intestinal obstruction. **Results:** Imaging studies revealed intussusception. He had mucocutaneous pigmentation and multiple hamartomatous polyps which were diagnosed histologically. The unusual presentation of the case and its successful management has prompted us to report the case with literature review.

Keywords: Peutz-Jeghers syndrome, intussusception, acute intestinal obstruction.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is an autosomal dominant disorder with variable penetrance characterized by the occurrence of gastrointestinal hamartomatous polyps in association with mucocutaneous hyperpigmentation and multiple neoplasms. This typically manifests itself as recurrent colicky abdominal pain and blood loss in stools. Hamartomatous polyps of Peutz-Jeghers syndrome grow to a large size and form the lead point of an intussusception. The diagnosis of is based on clinical findings and histopathological patterns of polyps.¹

CASE REPORT

A 19 yr old male presented with colicky abdominal pain in the periumbilical region associated with bilious vomiting and constipation of one day duration. Physical examination revealed multiple black coloured lesions over the buccal mucosa and both lips (Figure1), which the patient had since his childhood. Abdominal examination revealed tenderness in the umbilical and hypogastric regions associated with palpable distended bowel loops in the hypogastric region with diminished bowel sounds.

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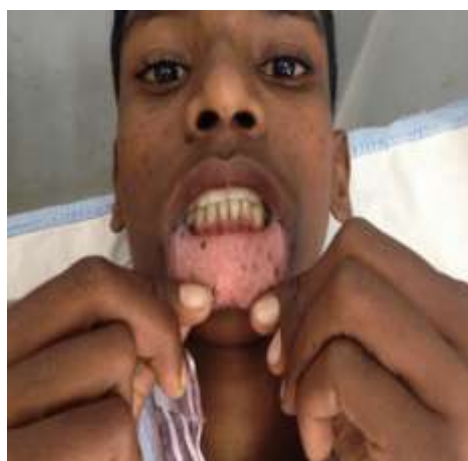


Figure 1

Mucocutaneous pigmentation

Laboratory investigations revealed anemia with Hb of 7.6gm% with associated polymorph nuclear leucocytosis (N-84%). Routine biochemical parameters were unremarkable. Radiograph showed dilated bowel loops with air fluid levels suggestive of acute intestinal obstruction. Ultrasonography (USG) revealed target sign with bowel in bowel with an echogenic portion between the loops revealing mesentery (Figure 2). Patient was adequately hydrated with intravenous fluids and two units of blood transfusion preoperatively. A provisional diagnosis of acute intestinal obstruction due to intussusception was considered.

The patient underwent an emergency exploratory laparotomy. Ileo-ileal intussusception was noted about 50 cms from ileocecal junction

and intussusception was reduced (Figure 3). A large polyp was seen in the lumen as the cause of intussusception (Figure 4). Small bowel segmental resection with end to end anastomosis was performed. The specimen was sent for histopathological examination (HPE). Post-operative course was uneventful.



Figure 2
USG showing intussusception



Figure 3
Intraoperative findings of ileo-ileal intussusception



Figure 4
Polyp causing intussusception

Histopathological examination revealed multiple sessile polyps (Figure 5) with one large polyp measuring 1.5 x 1 cm with dysplastic glands with nuclear atypia and increased mitotic activity and associated smooth muscles bundles (Figure 6) intersecting the glands. On the basis of hamartomatous small intestinal polyposis and clinical manifestation of mucocutaneous pigmentation, the diagnosis of Peutz-Jeghers syndrome was made.

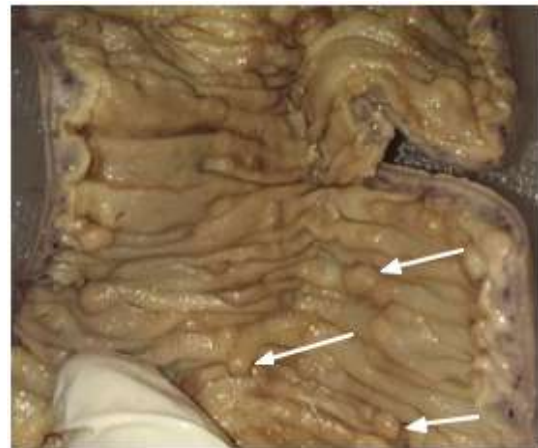


Figure 5
Ileum showing multiple polyps

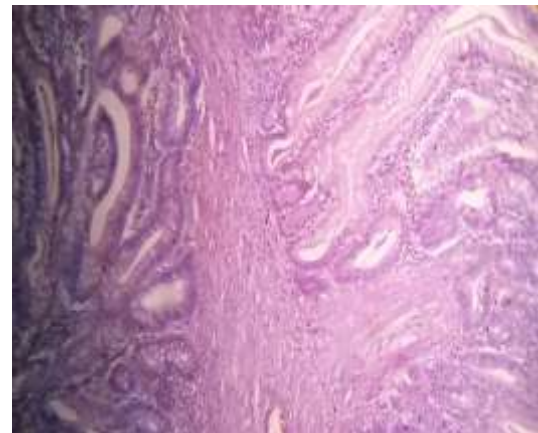


Figure 6
HPE Showing Smooth muscle bundles intersecting the glands

DISCUSSION

Peutz-Jeghers syndrome is a rare autosomal disorder with variable penetrance characterized by mucocutaneous hyper pigmentation, hamartomatous polyps of gastrointestinal tract and multiple neoplasms with both familial and sporadic transmission. The incidence of PJS is reported to be 1 in 8300 to 1 in 280,000 individuals. However, a probable prevalence of 1 in 100000 has been documented.² The gene associated with PJS is a serine-threonine kinase, the tumour suppressor gene; located on chromosome 19p13.3.¹ Hemminki et al and Jenne et al independently identified the gene in this region as LKB1/STK11

(serine/threonine-protein kinase 11, which is also known as LKB1) (3.4). This gene has been reported in 80% of patients with PJS. Up to 25% of recorded cases of PJS do not have family history. Those sporadic cases probably arise due to new mutation of STK11 gene or low penetration.

The median time to first presentation with polyps is about 11-13 years of age and approximately 50% will have experienced symptoms by the age of 20 years.⁵

The hyperpigmented lesions contain melanotic deposits and commonly manifest in infancy and childhood. The pigmented lesions are often seen on the lips, around the mouth, eyes, nostrils, on the buccal mucosa; and sparsely on the fingers, soles of the feet, palms, anal area and intestinal mucosa. Patients with PJS often present with a history of intermittent abdominal pain due to small bowel intussusception caused by the polyps. Some intussusceptions spontaneously reduce; others lead to development of small bowel obstruction. Peutz-Jeghers polyps can also ulcerate, leading to acute blood loss or chronic anemia. Although Peutz-Jeghers polyps are most commonly found in the gastrointestinal system, they can also occur in extra intestinal sites such as kidney, ureter, gallbladder, bronchial tree, nasal passages.⁶

Anemia, rectal bleeding, abdominal pain, obstruction, and intussusceptions are common complications in PJS. In adult, intussusception is rare and most commonly involves the small intestine, whereas in children, intussusception is commonly seen and the large intestine gets commonly involved.⁷ Intestinal obstruction (42.8%), abdominal pain (23%), rectal bleeding (13.5%) and extrusion of polyp (7%) are the common gastrointestinal presentations in PJS as found by Utsunomiya et al in their series of 222 patients with PJS. In their study they have shown that the most affected part of the gastrointestinal tract with polyps is small intestine (64%) followed by colon (63.2%), stomach (48.6%) and rectum (32%) (8). The incidence of polyps within the small intestine is the highest in the jejunum followed by ileum and duodenum.⁹ There are also reports of rare extraintestinal polyps, which include nasal polyps, gall bladder polyps, ureteric polyps and respiratory tract polyps.¹⁰

Those Individuals with PJS are at risk for the development of gastrointestinal & non-gastrointestinal malignancies. These patients have a 15-fold increased risk of developing intestinal cancer as compared with that of the general population.¹¹ Among the non-gastrointestinal type, pancreas, lung, breast, uterus, cervix, ovary, testis & thyroid are commonly involved.

The diagnostic criteria for the syndrome as proposed by Giardello et al are: histopathologically confirmed gastrointestinal polyps along with any two of the following three features: a) small bowel

polyposis, b) family history of PJS, c) pigmented macules of the buccal mucosa, lips, fingers and toes.¹¹

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. Adenomatous & carcinomatous changes have been described in Peutz-Jeghers polyps.¹²

The standard therapy for Peutz-Jeghers syndrome has been laparotomy and bowel resection to remove symptomatic gastrointestinal polyps that cause persistent or recurrent intussusceptions. Multiple surgical resections are sometimes required leading to short gut syndrome. Due to this reason, it has been recommended that endoscopy be performed to remove all the polyps. During each laparotomy, the small bowel should be examined by means of intraoperative enteroscopy (IOE). Nowadays, double balloon enteroscopy (DBE) in combination with capsule enteroscopy are the gold standard for the diagnosis and treatment of the small bowel hamartomatous polyps.²

Chemoprevention is under trial in order to reduce the polyp burden. The anti proliferative activity of macrolide immunosuppressant rapamycin (sirolimus) and the COX-2 inhibitor (celecoxib) are under investigational trial for chemo prevention in PJS.²

CONCLUSION

Peutz-Jeghers syndrome (PJS) is a rare syndrome. All the infants with pigmented macules in perioral regions should be investigated for PJS. There is no optimal screening strategy for PJS recommended so far. Hence, these patients should be kept for lifelong surveillance keeping in view the fact that they are more susceptible for intestinal as well as non intestinal malignancies.

REFERENCES

1. Mehenni H, Blouin JL, Radhakrishna U, Bhardwaj SS, Bhardwaj K, et al. Peutz-Jeghers syndrome: confirmation of linkage to chromosome 19p13.3 and identification of a potential second locus, on 19q13.4. *AmJHumGenet.* 1997; 61:1327-1334.
2. Marcela Kopacova, Ilja Tacheci, Stanislav Rejchrt, Jan Bures. Peutz-Jeghers syndrome: Diagnostic and therapeutic approach. *World J Gastroenterol.* 2009 November 21; 15(43): 5397-5408.
3. Hemminki A, Markie D, Tomlinson I, Avizienyte E, Roth S, et al. A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. *Nature.* 1998; 391:184-187.
4. Jenne DE, Reimann H, Nezu J, Friedel W, Loff S, et al. Peutz-Jeghers syndrome is caused by

- mutations in a novel serine threonine kinase. *Nat Genet.* 1998;18:38-43.
5. Vesna Živković et al. Hereditary Hamartomatous Gastrointestinal Polyposis Syndrome. *Scientific Journal of the Faculty of Medicine in Niš.* 2010;27(2):93-103
 6. Michael Manfredi. Hereditary Hamartomatous Polyposis Syndromes: Understanding the Disease Risks As Children Reach Adulthood. *Gastroenterology & Hepatology.* Volume 6, Issue 3 March 2010; 185-196.
 7. Hinds R, Philp C, Hyer W, Fell JM. Complications of child-hood Peutz-Jeghers syndrome: implications for pediatric screening. *J Pediatr Gastroenterol Nutr* 2004; 39(2):219-20.
 8. Utsunomiya J, Gocho H, Miyanaga T, Hamaguchi E, Ka-shimure A. Peutz-Jeghers syndrome: its natural course and management. *Johns Hopkins Med J.* 1975; 136(2):71-82.
 9. Gammon A, Jaspersen K, Kohlmann W, Burt RW. Hamar-tomatous polyposis syndromes. *Best Pract Res Clin Ga-stroenterol.* 2009; 23(2):219-31
 10. Cecilia M and Fenoglio-Preiser. Peutz-Jegher's syndrome. In: *Gastrointestinal Pathology: an atlas and text.* 3rd ed. Philadelphia: Lippincott Williams and Wilkins; pp.704-711, 2008.
 11. Giardello FM, Welsh SB, Hamilton SR et al. Increased risk of cancer in the Peutz-Jeghers syndrome. *N Engl J Med.* 1987;316:1511-1514.
 12. Karl H. Perzin, Mary F. Bridge, Adenomatous and Carcinomatous Changes in Hamartomatous Polyps of the Small Intestine (Peutz-Jeghers Syndrome): *Cancer.* 1982; 49: 971-983.

