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ACUTE ON CHRONIC INTESTINAL OBSTRUCTION OF A CASE OF PEUTZ-JEGHER SYNDROME - AN UNUSUAL PRESENTATION.

Gastroenterology

Das Kanhu Charan Professor, Department of Gastroenterology and hepatolgy, Apollo hospitals, Bhubaneswar.

Dr. Kailash Mohitey* Department of Gastroenterology and hepatolgy, Apollo hospitals, Bhubaneswar. *Corresponding Author

Dr. Ashish Barman Department of Gastroenterology and hepatolgy, Apollo hospitals, Bhubaneswar.

Dr. Pepse Pradhan Department of Gastroenterology and hepatolgy, Apollo hospitals, Bhubaneswar.

ABSTRACT

Peutz-Jeghers syndrome (PJS) is an autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition.Here We are going to present a case of Peutz-Jeghers syndrome 38 -year-old female presented with abdominal pain, vomiting & melena. The patient had pigmented lesions on the lower lip and buccal mucosa. The endoscopy revealed with multiple polyps involving stomach, duodenum , Colonscopy showed multiple small to medium size polys carped the colonic as well as visualized ileal mucosa.

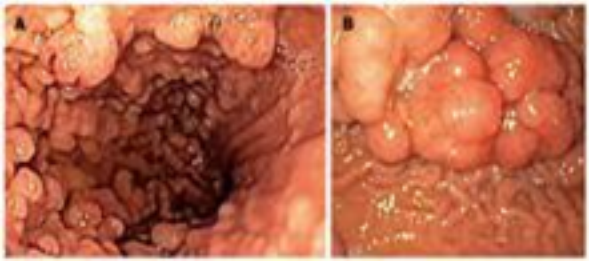
KEYWORDS

Peutz-Jeghers Syndrome, Mass abdomen, Recurrent small intestinal obstruction.

INTRODUCTION:
Peutz-Jeghers syndrome is an autosomal dominant inherited condition determined by a mutation localized at 19p13.3; characterized by the occurrence of gastrointestinal hamartomatous polyps in association with mucocutaneous hyperpigmentation. The diagnosis of PJS is based on clinical findings and histopathological patterns of polyps. PeutzJeghers syndrome is associated with significant morbidity, variable clinical course and considerable predisposition to gastrointestinal & non-gastrointestinal malignancies. An overall recommendation for PJS patients includes not only gastrointestinal multiple polyps resolution, but also regular lifelong cancer screening. Early detection and proper surveillance are vital to minimize the risk of carcinoma. Patients with Peutz-Jeghers syndrome have a 15-fold increased risk of developing intestinal cancer compared with the general population.

CASE REPORT:- A 38year-old female was admitted with sharp and intermittent abdominal pain, which was located in the periumbilical region. patient also reported nausea and several episodes of nonbilious, non-bloody vomiting. The patient had complained blackening of stool for five days. Physical examination revealed multiple, black coloured pigmented lesions on the face, lower lip, and buccal mucosa; which the patient had since childhood.

1)Pigmentations on the buccal mucosa & lower lip in a patient with PeutzJeghers syndrome.



Multiple pedunculated polyps in small intestine in this patient .

On abdominal examination, epigastric and periumbilical tenderness, diminished bowel sounds, and a possible epigastric mass were observed.

Laboratory investigations revealed low hemoglobin concentration: 8.2 g/dL with low MCHC . Routine blood chemistry reports were unremarkable. CECT showed multiple intraluminal filling defects carpeting the small bowel and large intestine suggestive of polypoid lesions in intestines. Spring coiled appearance of jejunum was noted in left upper abdomen, suggestive of intussusception with proximally dilated bowel loops. Abdominal ultrasonography (US) showed a large heterogeneous mass in the upper abdomen that measured 5.3 x 3.5 x 8.6 cm. The mass had a "pseudokidney" appearance with central, high echogenicity; consistent with mesenteric fat within an intussusception. The patient underwent an exploratory laparotomy. Small bowel segmental resection with end to end anastomosis was performed. The specimen was sent for histopathology examination histopathology revealed hamartomatous polyps . Post-operative course was uneventful.

DISCUSSION :
Peutz-Jeghers syndrome (PJS) is inherited disorder characterized by intestinal hamartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition. Here Peutz-Jeghers syndrome in a 38 -year-old female was a very rare case who presented to us abdominal pain, vomiting & melena. The patient had pigmented lesions on the lower lip and buccal mucosa. The endoscopy revealed with multiple polyps involving stomach, duodenum , Colonscopy showed multiple small to medium size polys carped the colonic as well as visualized ileal mucosa. This syndrome is also associated with extra-intestinal cancers like cancers of the breast, pancreas, testicles, ovaries, lung, cervix ect. However these type of lesion was not found in our patient.

CONCLUSION :
Such type of Young lady with features of recurrent intestinal obstruction having cutaneuos macular patches we should keep in mind of such syndrome. Patient should be investigated in that line to establish the diagnosis. Family screening should be advised for early detection. Genetic testing of the sibling should be carried out if possible because most of the time this is diagnosed with help of clinical findings and endoscopic –histological test.

Competing interests:
The authors declare that they have no competing interests.

Authors' contribution: *Dr. Kailash Mohitey , Dr. Ashish Barman. Dr.P. Pradhan* were involved in the clinical assessment and writing the case report. All authors read and approved the final manuscript.

Consent:
Full written consent was received for the manuscript to be published.

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