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Case Report

Peutz-Jeghers Syndrome: A Rare Case Report

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Abstract

We report a case of 26-year-old male presenting with features of intestinal obstruction which on ultrasonography was S/O intussusception. On examination, patient was having distended abdomen with generalized tenderness and guarding and multiple pigmented spots over his face, arms, and other parts of the body. On exploratory laparotomy, intussusception was found with ileum and jejunum as intussusceptum and colon as intussuscepien. On resection of jejunum and ileum multiple polyps were found which on histopathological examination were found to be hamartomatous polyps. Upper giscopy also showed polyps in different parts of stomach. These polyps with melanoma cutaneous pigmentation suggest a diagnosis of Peutz-Jeghers syndrome. This syndrome is a rare entity with patients having increased chances of intestinal and extra intestinal malignancy and hence should be subjected to regular follow-up with screening for other malignancies.

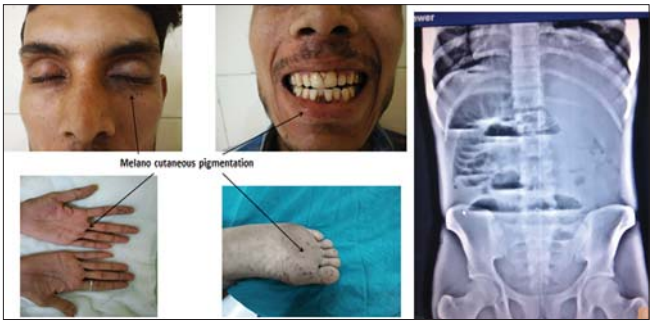
Key words: Hamartomatous polyp, Melanoma cutaneous pigmentation, Peutz-Jeghers syndrome

INTRODUCTION

Peutz-jeghers is autosomal dominant syndrome characterized by hamartomatous gastrointestinal (GI) polyps (<100) and mucocutaneous melanin pigmentation. These polyps are 1 mm to 4 cm in diameter, mostly seen in jejunum and small bowel > colon. They can also occur at nose, bronchi, renal pelvis, and biliary tree.[1-3]

CASE REPORT

A case of 26-year-old male patient, presented with complain of severe colicky pain in whole of abdomen since 7-8 days associated with multiple episodes of vomiting and passing blood in stool for 2 days. There was no significant last history. His father suffered from GI malignancy and died in his forties, but the details of the malignancy were not available. Patient was having pallor and was malnourished (weight: 40 kg). On gross inspection, multiple pigmented spots present on face and digits of arms and legs, on bucco-oral mucosa, and on lips. Per abdomen was distended with generalized tenderness and guarding. There was a lump of 6 cm × 6 cm size present in left lumbar region extending into the umbilical region. On Per rectum examination stool mixed with blood with normal mucosa, and normal prostate size was found. Proctoscopy was normal with no visible pigmentation.



Blood investigations and X-ray chest were normal. X-ray abdomen standing was S/O multiple air fluid levels. Ultrasonography was S/O clumped small bowel loops in left hypochondrium and epigastric region P/O intussusception.

Intraoperative Findings

On opening peritoneum, jejuno-ileal type of intussusception was found which was reduced intussucipiens was found to be jejunum and some part of ileum and intussuceptum was distal ileum and colon. Most of the intussucipiens

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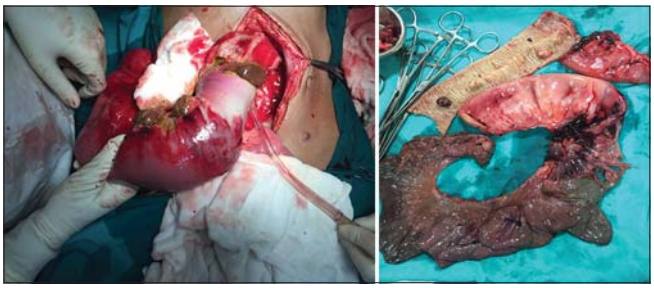
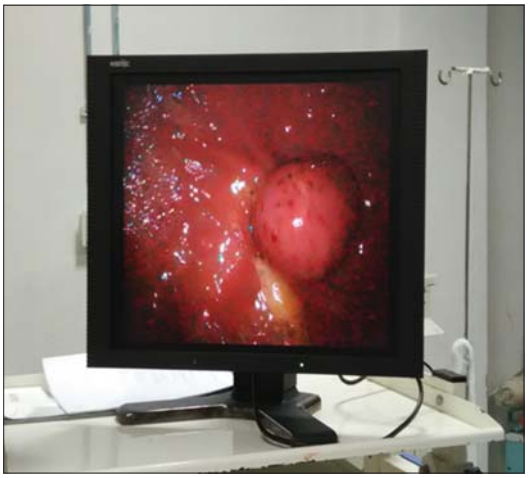
was gangrenous and sloughed out and was resected with jejuno-ileal anastomosis being done. Resected part had a number of polyps in its lumen which on histopathological examination was S/O hamartomatous polyps.



Hence postoperatively upper giscopy was done which revealed polyps in the distal part of stomach. Colonoscopy is awaited in this patient.



DISCUSSION

Peutz-jeghers is autosomal dominant syndrome characterized by hamartomatous GI polyps (<100) and mucocutaneous melanin pigmentation. These polyps are 1 mm to 4 cm in diameter, mostly seen in jejunum and small bowel > colon. They can also occur at the nose, bronchi, renal pelvis, and biliary tree. Pigmentation is seen in perioral, buccal mucosa, digits of hands and feet and perianal and genital region. These patients mostly present in 3rd decade with abdominal pain due to intussusception. Other less common presentations: Symptoms from obstruction of large polyps, anemia, hematochezia, hematemesis, biliary obstruction, and gastric outlet obstruction. They are at an increased risk of both GI and extra GI malignancy such as that of pancreas, breast, thyroid, lungs, gallbladder, ovary, and testes.[3-6]

CONCLUSION

Because of the increased malignancy chances, the patient is kept on regular follow-up with screening with intervention as and when required.

Screening Guidelines

Colon and rectum	Baseline colonoscopy at age 8
	If polyps detected, colonoscopy every 3 years until age 50
	If no polyps detected, repeat colonoscopy at age 18 and every 3 years after until age 50
	Continue surveillance at 1- to 2-year intervals after age 50
Small intestine	Baseline EGD at age 8
	If polyps detected, EGD every 3 years until age 50
	If no polyps detected, repeat EGD at age 18 and every 3 years after until age 50
	Continue surveillance at 1- to 2-year intervals after age 50
	Baseline video capsule endoscopy at age 8
	Repeat every 3 years
Genital tract	Annual testicular examination beginning at birth until 12 years
	Testicular ultrasound if abnormalities on examination
	Cervical smear with liquid-based cytology at age 25 years
	Repeat every 3 years
Breast	Monthly self-examination beginning at age 18
	Annual breast MRI from age 25 to 50
	Annual mammography beginning at age 50
General	Annual complete blood count and liver function tests
	Annual full physical examination

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