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CASE REPORT

Peutz-Jeghers Syndrome presenting as Intussusception in a young female: A Case Report

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ABSTRACT

Peutz-Jeghers syndrome (PJS) is rare condition with autosomal dominant inheritance. It is due to mutation in the tumor suppressor gene, serine threonine kinase 11 (STK11/LKB1) at chromosome 9. It has a very characteristic presentation. Patient usually presents at a young age with intestinal obstruction due to polyps leading to intussusception. There is also melanosis on digits, perioral and perineal regions. There is a very strong association of cervical and breast carcinoma in females and testicular tumors in males. GIT, pancreatic and lung malignancies occur in both sexes. Regular follow up and screening are necessary for early detection of malignancies. We present a case of a young girl, who presented with intestinal obstruction along with other signs and symptoms of Peutz-Jeghers syndrome. Exploratory laparotomy revealed jejunoileal intussusception and hamartomatous polyps. Patient and other family members were advised screening studies of associated malignancies and regular follow up.

Key Words: Hamartomatous polyps, Intussusception, Melanosis, Peutz-Jeghers syndrome

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Introduction

Peutz-Jeghers syndrome is an uncommon entity occurring only 1 in 8300 to 1 in 280,000 births.[1] It is an autosomal dominant disease characterized by hamartomatous polyps in jejunum, ileum and colon. There is melanosis of mucocutaneous junctions such as in the mouth and perineal regions. Melanin spots also appear on the digits.[2]The polyps are rarely premalignant, but colorectal cancer and malignancies of breast and cervix are strongly associated with this syndrome. Lungs, pancreas, uterus, ovaries and testis may also undergo malignant transformation.[3]

Polyps serve as pathological lead point and patient presents with acute intestinal obstruction due to intussusception at an early age. Intussusception and gastrointestinal bleeding are common presentations, which are commonly treated by resection of the diseased bowel.[2] Regular surveillances for associated cancers and long-term follow-up is necessary to decrease mortality and morbidity.

Here we report case of a young girl who presented with intestinal obstruction and jejunoileal intussusception due to multiple polyps. She had melanosis around the mouth and inside the buccal mucosa and on the digits. She was diagnosed as a case of Peutz-Jeghers syndrome.

Case Report

A 15-year-old female resident of Dera Ismail Khan, Khyber Pakhtunkhwa (KPK) presented at emergency department of Nishtar Medical University and Hospital, Multan with

complaints of generalized abdominal pain which was acute in onset, vomiting for 3 days, constipation and abdominal distention for 02 days. Patient gave history about the presence of brown spots around the mouth and fingers since childhood. Two other siblings were also having such spots. She did not give history of fever, weight loss or previous surgery. There was no previous history of pulmonary tuberculosis. Patient was a non-smoker and not addicted to anything. Her eating habits were non-significant.

Her clinical examination revealed a lean female of average height, with melanin spots around the mouth (Figure 1A) and on the fingers (Figure 1B). Heart rate was 108/minute, blood pressure 100/60 mmHg and respiratory rate 28/minute. On palpation, abdomen was distended, tense and tender with intact hernial orifices. Lymph nodes were not palpable. Bowel sounds were absent. Digital rectal examination was unremarkable. CNS, respiratory system and cardiovascular examination was also unremarkable.

Her hemoglobin was 13.9 g/dl and total leukocyte count was 15,490/mm^3. Renal and liver function tests were normal. Abdominal supine X-ray showed valvulae conniventes indicating small bowel obstruction. Ultrasound abdomen revealed aperistaltic gut loops. Chest X ray was unremarkable.

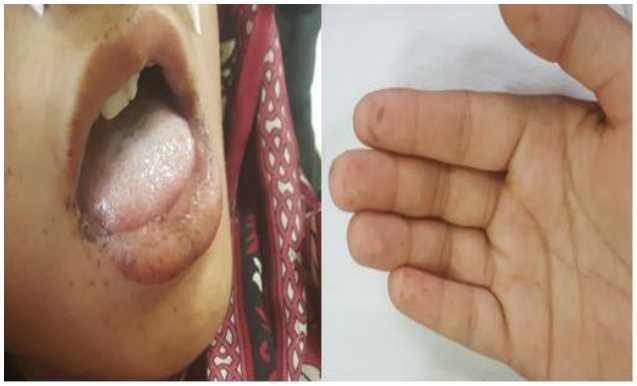

Figure 1: Patient with Peutz-Jeghers syndrome exhibiting circumoral melanin spots (A) and digital melanosis (B)

Exploratory laparotomy revealed jejunoileal intussusception leading to jejunal dilatation and small bowel obstruction. About 75cm of the diseased portion of jejunum was resected and primary anastomosis done. The diseased portion was having six hamartomatous polyps largest one was approximately 3cm in size and the smallest was of 1 cm as shown in Figure 2. Patient was kept nil per oral and on intravenous fluids for four days. She was discharged on fifth postoperative day. Her postoperative follow-up visit was uneventful. She was advised ultrasound breast, CT scan abdomen and referred to gynecologist to screen for associated cervical malignancy. The other family members were also advised to go for the screening studies for early detection and management of anticipated pathologies.

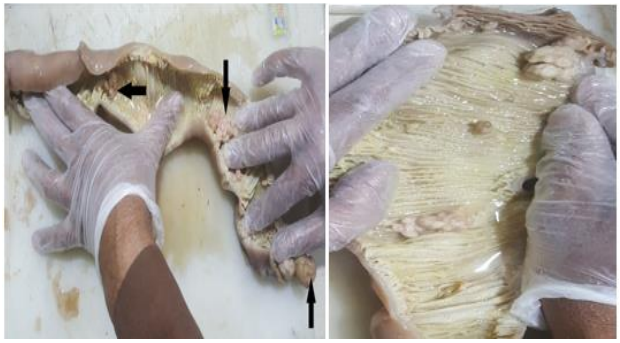

Figure 2: Gross specimen of resected portion of jejunum showing hamartomatous polyps

Discussion

Peutz-Jeghers is an uncommon autosomal dominant syndrome which presents with very typical signs and symptoms and mutation of tumor suppressor gene, STK11/LKB1 on chromosome 9.[4] Melanin pigmentation first appears during infancy and increases until puberty. This pigmentation appears at buccal mucosa, vermillion border of the lip, tip of fingers, toes and perianal region. Usually melanosis at buccal mucosa persists throughout life while melanin spots in other areas fade away. Vermillion border of the lip is most commonly involved, followed by the buccal mucosa, fingers and toes.[5] Melanin spots were present on the vermillion border of the lips, fingers and toes in this patient (Figure 1).

The following WHO criteria as outlined by Riegert-Johnson et al[6] were used for confirming the diagnosis of Peutz-Jeghers syndrome:

1. Any number of hamartomatous polyps,
2. Family history of PJS,
3. Prominent melanosis at mucocutaneous junctions

In our patient there were multiple hamartomatous polyps, pigmentation of mucocutaneous junctions and a positive family history of PJS.

Patients with PJS usually presents with intestinal obstruction due to intussusception. Hamartomatous polyps are not premalignant and are mostly located in small intestine, colon and stomach.[7] The life-time risk of gastrointestinal and non-gastrointestinal cancers is high and strict surveillance is necessary for early detection.[8] Our patient presented with intestinal obstruction due to jejunoileal intussuseption without any malignancy. Screening for GIT malignancy was previously done by barium studies and enteroclysis. However newer technologies such as gastroscopy, colonoscopy, double balloon enteroscopy and capsule endoscopy have now taken the role of surveillance tools for gastrointestinal pathologies.[8]

According to a study by Kastrinos and colleagues, pancreatic carcinoma is a common cancer associated with PJS with 36% risk. Regular screening of high-risk patients is done by CT scan and more reliably by endoscopic ultrasound on yearly basis.[9] Pap smear, transvaginal USG and mammography are also advised on a yearly basis[2]. We discharged our patient with the advice of regular follow-up and screening. Family of the patient was also counseled and advised about the screening of other two siblings with melanin spots.

Conclusion

Peutz-jeghers syndrome is a rare autosomal dominant disorder with melanosis (circumoral, perianal and digital) and hamartomatous polyps (intestinal) as characteristic features of this disease. Regular screening of all the family members is necessary due to a very strong association of breast, cervical, colonic and other cancers as early detection of these associated malignancies can save many lives.

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