ORIGINAL RESEARCH PAPER

General Surgery

KEY WORDS:

A RARE CASE REPORT OF PEUTZ JEGHERS SYNDROME

Dr. Hiral .C. Chauhan Assistant Professor, Department of General Surgery, B.J.Medical College, Civil Hospital, Ahmedabad.

Dr. Rinav. M. Kenia* 3rd Year Resident, Department Of General Surgery, B.J.Medical College, Civil Hospital, Ahmedabad. * Corresponding Author

Dr. Nina .M. Shah Associate Professor And HOU, Department of General Surgery, B.J.Medical College, Civil Hospital, Ahmedabad.

INTRODUCTION

Peutz–Jeghers syndrome (often abbreviated **PJS**) is an autosomal dominant genetic disorder characterized by the development of benign hamartomatous polyps in the gastrointestinal tract and hyperpigmented macules on the lips and oral mucosa (melanosis). This syndrome can be classed as one of various hereditary intestinal polyposis syndromes and one of various hamartomatous polyposis syndromes. It has an incidence of approximately 1 in 25,000 to 300,000 births. Peutz-Jeghers syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to increase the risk of developing noncancerous polyps and cancerous tumors. In about half of all cases, an affected person inherits a mutation in the *STK11* gene from one affected parent. The remaining cases occur in people with no history of Peutz-Jeghers syndrome in their family. These cases appear to result from new (de novo) mutations in the *STK11* gene.

CASE REPORT

This is a case report of a 13 year old male patient who presented with complaint of generalized abdominal pain which was vague and poorly localized; generalized distention with bilious vomiting and not passing stool since last 2 days. On examination there is generalized abdominal distention with tenderness in periumbilical region. Bowel sounds was hyperperistaltic.

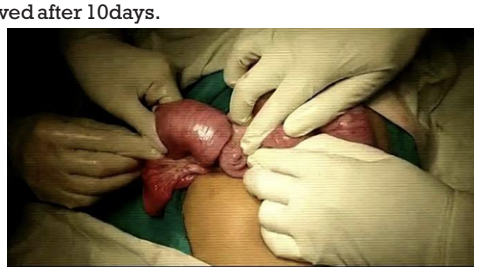
On per-rectal examination ballooning was present.

Abdomen Xray was suggestive of dilated small bowel loops with multiple air fluid levels. Ultrasound of abdomen was suggestive of jejunoileal intususscception with dilated and content filled small bowel loops of average diameter 30mm with to and fro movement.

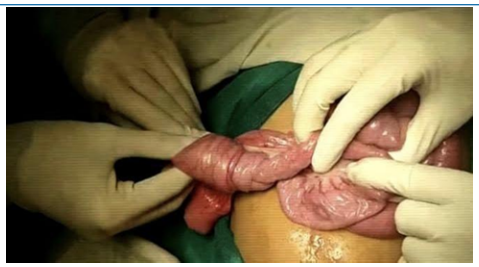
Emergency exploratory laparotomy was done.

PROCEDURE

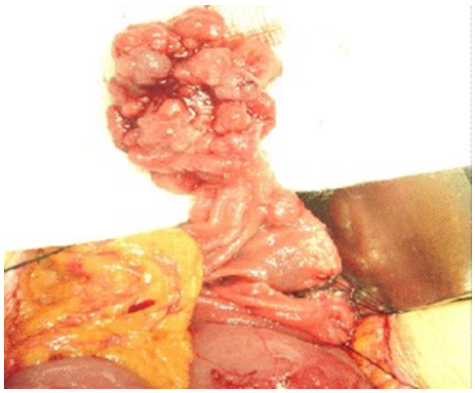
Under general anesthesia and maintaining aseptic precautions, a vertical mid-midline incision was made. On opening the peritoneum , an ileo-ileal intussucception was identified and was reduced. Two polypoidal mass of approximately 2x2cm and 2x1cm was found 3cm apart in the ileum as the lead point which were excised with an enterotomy and sent for histopathological examination. Rest of the bowel loops were screened for other polypoid lesions and were found to be normal. The abdomen was closed in layers. Post-operative period was uneventful and sutures were removed after 10days.



Peroperative intussusception

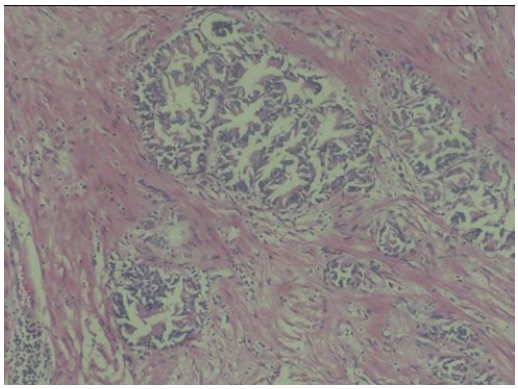


Partially reduced intussusception



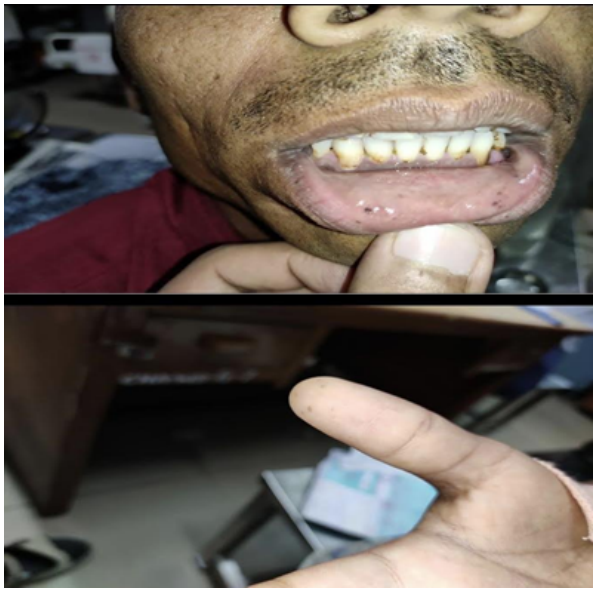
Intraoperative finding of polyp upon enterotomy.

Histopathological examination was showing structure of polyp with evidence of disorganized glands admixed with intervening ramifying bundles of smooth muscle cells and no evidence of dysplasia; findings suggestive of Peutz Jeghers Polyp.

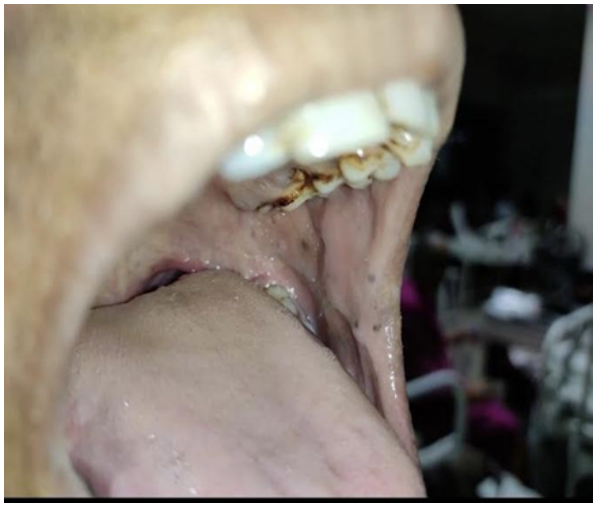


Histopathological finding of the polyp

Upon retrospective examination, the patient had hyperpigmented macules on the lips,oral mucosa and finger tips consistent with presentation of peutz jeghers syndrome. Patient's father also had hyperpigmented macules on the lips and oral mucosa.



Hyperpigmented macules in oral mucosa and palm of father.



Hyperpigmented macules in patient's oral cavity

DISCUSSION

Peutz–Jeghers syndrome (PJS) is an autosomal dominant disorder due to mutation of STK11 gene on chromosome 19p which codes for serine-threonine kinase. Clinically, it presents with hundreds of hamartomatous polyps in the gastrointestinal tract along with mucocutaneous involvement characterized by hyperpigmented macules on the buccal mucosa, lips, periorbital area, nose, back of the hands and tips of fingers and toes, and rarely over palms and soles.These mucocutaneous lesions appear in infancy and early childhood and generally disappear post- puberty with the oral lesions persisting lifelong.

The clinicopathological criteria of the World Health Organisation for diagnosing PJS are as follows:(any one criteria is sufficient for the diagnosis of PJS)

1. Three or more polyps, with histological features of PJS
2. A family history of PJS with any number of polyps
3. A family history of PJS with characteristic mucocutaneous pigmentation

The hamartomatous polyps most commonly occur in the small intestine (78%) followed by colon (42%), stomach (38%), and rectum (28%). Maximum number is found in jejunum followed by ileum and least in the duodenum. They may occur at any other part of the gastrointestinal tract or even outside the gut, like renal pelvis, urinary bladder, ureters, lungs, nares, and gallbladder. Rarely, polyps may be adenomatous which might create confusion with familial adenomatous polyposis syndrome.There is an increased risk of intestinal and nonintestinal malignancies associated with PJS. Colorectal cancer is the most common among the gastrointestinal malignancies. Gastric and pancreatic cancers are less common than colorectal. Extraintestinal malignancies include cancer of the breast, ovary, cervix, fallopian tube, thyroid, lung, gallbladder, bile duct, pancreas, and testis.

The polyps are generally benign but can lead to an emergency situation due to a small bowel obstruction (intussusception being the most common cause), rectal prolapse and excessive hemorrhage leading to anemia and heart failure. The more common types of intussusception are jejunojejunal, jejunoileal, ileoileal, ileocecal,ileocolic and colocolic type. The patient generally presents to the emergency with features of acute abdomen, i.e., pain, constipation/obstipation, nausea, vomiting, abdominal distention, tachycardia, tachypnea, and hypotension. After the initial resuscitation, emergency X-ray abdomen , Ultrasound abdomen and if indicated, computed tomography (CT) abdomen is advisable. Following confirmation, exploratory laparotomy should be done.

An early suspicion, detection and proper surveillance of PJS can reduce the incidence of this dramatic and at times, fatal outcome. The patients generally present with hyperpigmentedmacules at around 5 year of age and positive family history of PJS can be elicited. In most cases, the other features of PJS are not evident at this age. If an early diagnosis can be established at this age and surveillance done according to the guidelines, there are two advantages. First, early detection of any complication and malignancy would be possible. Second, an elective surgery can be planned for prophylactic polypectomy such as double balloon enteroscopy and intraoperative enteroscopy. Some consider these polyps as premalignant lesions, which shouldbe removed without delay, by surgery or endoscopy. However, others state that the rate of malignant transformation in these polyps are very low, so periodic endoscopic surveillance is sufficient.

CONCLUSION

Peutz-Jeghers syndrome should be paid more attention because of the risk of complications related to polyps, and the association with cancers. Peutz-Jeghers syndrome patients should have a periodical assessment of the ailments of digestive tract.

REFERENCES

1. Dormandy TL. Gastrointestinal polyposis with mucocutaneous pigmentation(Peutz-Jeghers syndrome) N Engl J Med. 1949;241:993–1005. [PubMed] [GoogleScholar]
2. Tsao H. Update on familial cancer syndromes and the skin. J Am AcadDermatol. 2000;42:939–69. [PubMed] [GoogleScholar]
3. Shivaswamy KN, Shyamprasad AL, Sumathy TK, Ranganathan C. Peutz-Jeghers syndrome with prominent palmoplantar pigmentation. Indian J Dermatol Venereol Leprol. 2008;74:154–5. [PubMed] [GoogleScholar]
4. Mozaffari HR, Rezaei F, Sharifi R, Mirbahari SG. Seven-year follow-up of Peutz-Jeghers syndrome. [Last accessed on 2016 Sep 03];Case Rep Dent. 2016 2016:6052181. Availablefrom: https://ncbi.nlm.nih.gov/pubmed27195155 . [PMC free article] [PubMed] [GoogleScholar]
5. Lynch HT, Lynch JF, Lynch PM, Attard T. Hereditary colorectal cancer syndromes: Molecular genetics, genetic counseling, diagnosis and management. Fam Cancer. 2008;7:27–39. [PubMed] [GoogleScholar]
6. Hearle N, Schumacher V, Menko FH, Olschwang S, Boardman LA, Gille JJ, et al.Frequency and spectrum of cancers in the Peutz-Jeghers syndrome. Clin Cancer Res. 2006;12:3209–15. [PubMed] [GoogleScholar]
7. Ozer A, Sarkut P, Ozturk E, Yilmazlar T. Jejunoduodenal intussusception caused by a solitary polyp in a woman with Peutz-Jeghers syndrome: A case report. J Med Case Rep. 2014;8:13. [PMC free article] [PubMed] [GoogleScholar]