Case Report

PEUTZ-JEGHERS SYNDROME IN A GIRL WITH CHRONIC ABDOMINAL PAIN

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ABSTRACT
It is a report of a seven years old girl with Peutz-Jeghers syndrome. Mode of presentation, various investigations, pattern of family history and management aspect are reported.

Key Words: Peutz Jeghers, Rare syndrome, Strong familial association.

INTRODUCTION
Peutz-Jeghers syndrome is an autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps in association with mucocutaneous melanotic pigmentation. Historically it was first describe by Peutz in 1921, although Hutchinson had described the distinctive pigmented spots of the lips in 1896.

However, he was unaware of association of intestinal polyp. In 1949 Jeghers established this association in 22 cases.1

Recently incidence of malignancy has been noted by several authorities in literature. Further investigations has established a from of mutation of the LKB1/STK11 gene, located on chromosome 19p13.3.2-4 From polyposis registries it would appear that Peutz-jeghers syndrome is rare and is only 10% compared to Familial adenomatous polyposis. This condition has been manifest from two to 82 years of age with in average of 29 years. Incidence is almost equal in male and females. Pigmented lesions are usually present in the first few years of life and may fade at puberty, except for those which are on the buccal mucosa. This may provide a clue to diagnosis in pediatric patients. Therefore a high index of suspension is required.

CASE REPORT
A seven years old girl who presented with a past history of crampy abdominal pain from the age of two years and circumoral pigmentation was noted at that time (Fig-1). The pigmentation increased by the time she was four
At that time a colonic polyp was removed through colonoscopy. On this occasion we performed upper gastro intestinal endoscopy and removed two pedunculated polyps from proximal jejunum. However no polyps were seen on colonoscopy. Her family history was significant because her father was discovered to have ulcerating gastric cancer. She has been put on the list of regular follow up.

**DISCUSSION**

About one third of patient with Peutz Jeghers syndrome will have symptoms like crampy abdominal pain, abdominal mass due to intussusception signs of intestinal obstruction in their first decade of life and 50 to 60% present before the age of 20 years. Often they are anemic. The small and large bowel polyps in these patients tend to be pedunculated where as stomach polyps are generally sessile. These polyps can attain a very large size and these pedunculated polyps are generally lead point of intussusception. A case has been reported of 24 year old man with Peutz Jeghers syndrome who developed small intestinal adenocarcinoma and presented with small bowel obstruction due to jejunoleal intussusception. 

Interestingly intralobular cell hyalinizing sertoli cell neoplasia of the testis was reported in eight cases with this syndrome. Moreover 5% of females have been seen to develop a peculiar ovarian tumor, which is called sex cord tumor with annular tubules. There has been recommendations that esophagogastroduodenoscopy, colonoscopy, upper gastrointestinal series with small bowel follow through should be carried out in patients with Peutz-Jeghers syndrome, starting at the age of 10 years and every two years thereafter, although in some cases follow-up radiologic and endoscopic examination are based on clinical course and symptoms.

In conclusion it is important to be vigilant and observant in patients who have circumovral pigmentation to go into the depth of history and physical findings and where indicated endoscopic procedures should be performed.

**REFERENCES**