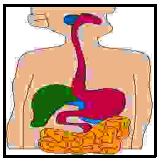

CASE REPORT PROF-576

PEUTZ JEGHERS SYNDROME;
THE POLYP MAY BE MALIGNANT

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ABSTRACT

Peutz-Jegher's syndrome is an uncommon disease. A case of 9 years old girl is presented who had signs and symptoms of subacute intestinal obstruction. She had mucocutaneous pigmentation of mouth and lips as well. Laparotomy revealed irreducible ileoileal intussusception with a polyp as lead point. Histopathological examination of the polyp showed malignant change. The polyps of Peutz-Jegher's syndrome had the reputation of being hamartomas but lately, cases have been reported of malignant changes in them. Literature has been reviewed to reassess the management of these cases due to increasing evidence of malignancy.

KEY WORDS: Peutz-Jegher's Syndrome, Malignant , Polyp

INTRODUCTION

Peutz-Jegher's Syndrome (PJS) is a rare disease characterized by mucocutaneous pigmentation associated with polyposis of gastrointestinal tract. The classical pigmentation of PJS distinguishes it from other polyposis syndromes[1].

The nature of polyps remained controversial for a long time but are now considered to be hamartomas[2]. Intestinal tumors associated with PJS and malignant changes in the hamartomatous polyps have been reported in the past. The polyp causing intussusception in the reported case also developed malignant change . These tumors need to be dealt aggressively and a well planned follow up is required for these patients.

CASE REPORT

A 9 years old girl was admitted through out patient department with history of abdominal pain of one month's duration which was diffuse and continuous. It was associated with off and on vomiting, constipation and abdominal distension. On examination her pulse was 100/min, blood pressure, temperature and respiratory rate were normal. She had blackish brown pigmentation on her lips and buccal mucosa (Fig 1).

Her abdomen was tender and a 9x12 cm mass was palpable in the umbilical region. Rectal examination was inconclusive. Laboratory investigations revealed hemoglobin of 10g/dl whereas TLC, DLC and ESR were normal. Serum sodium and potassium were also within normal range. Plain x-rays of the abdomen in erect posture showed multiple air fluid levels with distended small bowel loops. Ultrasonography of the abdomen revealed signs suggestive of intussusception. Diagnosis of Peutz Jegher's syndrome with intussusception was made. The patient was prepared for surgery after resuscitation. Laparotomy showed

ileoileal intussusception about 60 cm from ileocecal junction with a polyp as lead point.

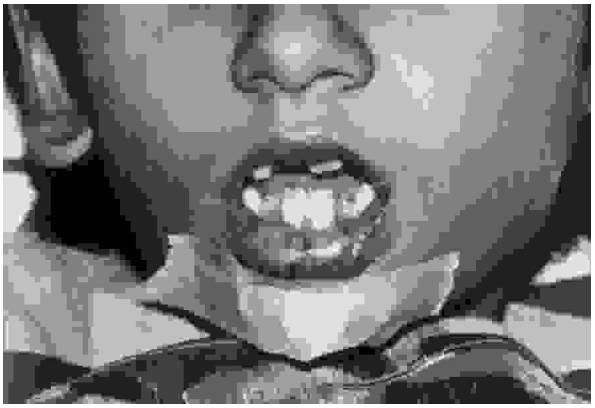

Figure-1

The intussusception was irreducible, so resection with end to end anastomosis had to be done. Palpation of the intestines revealed three other polyps, one was at about 15 cm from duodenojejunal flexure whereas the second was about 45 cm distal to the first one. Third polyp was found in mid jejunum (Fig 2).

Figure-2

All these polyps were excised through enterotomies. Mesenteric lymph nodes appeared normal and a few were excised for histopathological examination. The child made an uneventful recovery. Histopathological examination of polyp causing intussusception revealed malignancy i.e. adenocarcinoma. The margins of resected intestine, lymph nodes and other polyps were clear of any malignancy. However the case was referred to oncology department for further management.

DISCUSSION

Intestinal polypsis associated with mucocutaneous pigmentation was first noted by Peutz in 1921 and later established as a definite entity by Jegher et al in 1949[3]. It is inherited in an autosomal dominant pattern but some may develop de novo, most likely representing new, spontaneous mutations[4]. The disease has an equal sex distribution. The hallmark of the syndrome is intestinal polyposis associated with pigmented spots on buccal mucosa, lips and skin of face, hands and feet. The polyps in PJS are hamartoma, which principally occur in small intestine (55%) but can be found in colon and rectum (15%) or stomach and duodenum (30%)[5].

The polyps in PJS are usually multiple, lobulated hemispherical protrusions with a pedicle. Small sessile growths may also occur. Presenting features are usually repeated attacks of colicky abdominal pain caused by intussusception. Frank bleeding per rectum may also occur. Anaemia due to occult or obvious blood loss may be present. Mucocutaneous pigmentation may sometimes be the initial sign. Common bile duct obstruction and pancreatitis due to intussusception caused by a polyp in duodenum has also been reported[6]. Diagnosis is usually made clinically but barium studies and endoscopy of upper and lower alimentary tract are helpful diagnostic tools of PJS.

Although polyps of PJS have enjoyed the reputation of being benign lesions, malignant transformation can occur in them. This may be due to hamartoma-adenoma-carcinoma sequence occurring in small intestine of patients with PJS. Areas typical of hamartoma as well as hyperplasia, adenoma with mild to moderate dysplasia and carcinoma in situ all in the same polyp seen in a case of PJS suggest that hamartomatous polyp of PJS has the potential to undergo malignant transformation[7]. Cancerous transformation of polyp starts with stalk invasion. No statistical difference in size of hamartomatous and neoplastic polyps are seen[8]. Patients of PJS have increased risk of developing intestinal and extra intestinal malignancies. At least 1/3 of these patients are expected to develop cancer of gastrointestinal tract. Patients with PJS have a relative risk for death from gastrointestinal cancers that is 13 times greater and from all cancers 9 times greater when

compared with an age-matched general population[4]. Extra intestinal tumours associated with PJS include ovarian, cervical and testicular tumours. Cancer of breast, thyroid, bile duct, pancreas and gall bladder are also associated with PJS[4].

Since PJS is now recognized as a cancer predisposing syndrome, the treatment has changed over the past decade. The conventional treatment of PJS is conservative unless some complication of the disease like intussusception or hemorrhage takes place. Since polyps occur in crops, it is not possible to avoid recurrence . Extensive intestinal resection with repeated surgery can lead to short bowel syndrome[9].

The management protocol currently being followed is that all polyps larger than 0.5 cm in diameter found at endoscopy should be removed[4]. Combined surgical and endoscopic approach offers accurate assessment of extent of polyposis with removal of small polyps endoscopically. Endoscopy can also direct the surgeon to select the enterotomy site for excision of larger polyps. It is not very extensive surgical resection of intestine and threat of short bowel syndrome can be avoided[10,11]. Per operative enteroscopy decreases the re-laparotomy rate[12]. Further management of patients with PJS is based on the fact of increased risk of developing malignancy in patients with PJS. This includes annual assessment of the patient for (a) symptoms related to polyps, (b) breast and pelvic ultrasonic examination in girls (c) testicular examination in boys (d) pancreatic ultrasonography. Biennial enteroscopy along with gastrointestinal contrast studies. Mammography is advised at 25, 35 and 38 years of age, biennially until 50 years of age and then annually.

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