Case Report

NEEDLESS PAIN- RECURRENT INTUSUCCPTION: A SEQUELAE OF PEUTZ JEGHERS SYNDROME. RARE CASE REPORT AND REVIEW OF LITERATURE

Kumar,S, Ambikavathy,M

* Associate professor, Dept of Medicine, Vydehi Institute Of Medical Sciences and Research Centre, whitefield, Bangalore-66
* Associate professor, Dept of General Surgery, Vydehi Institute Of Medical Sciences and Research Centre, whitefield, Bangalore-66

1. Introduction

Peutz-Jeghers syndrome is a condition in which multiple hamartomatous polyps are present in the gastrointestinal tract in association with distinctive mucocutaneous pigmentation. Males and females are equally affected and can occur in any racial or ethnic group. It is considered rare with prevalence ranging between 1:250,000 and 1:300,000 and in India 4,260 per 1,065,070,607 population. Patients with Peutz-Jeghers syndrome often present with history of pain abdomen due to small bowel obstruction/intussusception. We report a case of a male patient aged 22 years who presented with severe anaemia and chronic pain abdomen with bleeding per rectum from one year on and off. The patient was diagnosed as Peutz-Jeghers Syndrome with sub acute small bowel obstruction due to recurrent intussusceptions. He underwent laparotomy with reduction of intussusceptions and resection and anastomosis along with intraoperative enteroscopic polypectomy. Histopathological examination confirmed as hamartomatous polyp of small bowel and colon. The patient was symptom free at follow up at six months post surgery, and he was lost to follow up after that.

2. CASE ILLUSTRATION:

A 22 years old patient was brought to Medical out patient department with history of generalised weakness, pain abdomen and passing blood in stools for a period of one year. He gave history of vomiting at the time of pain abdomen, followed by relief.

On physical examination he had characteristic melanotic brownish pigmented spots over the vermilion border of lower lip and buccal mucosa. Periorbital pigmentation was faintly seen. The patient was severely anaemic.

On Per abdomen examination mild distension was noted. Palpation was tender in the periumbilical region with a palpable mass in the epigastric region measuring about 3x5x6cms. Bowel sound was sluggish. Per rectal/proctoscopy revealed mucosal prolapse and a rectal polyp. There was a family history of his uncle who died of similar complaints.

A clinical diagnosis of Peutz-Jeghers syndrome with sub acute small bowel obstruction secondary to intussusception with severe anaemia was made and patient was subjected to various investigations.

The initial erect x-ray abdomen demonstrated a nonspecific bowel gas pattern with no evidence of bowel obstruction or free air. Ultrasoundography showed a heterogenous mass in the epigastric region measuring 4x5x4x6cms giving an appearance of a "pseudo kidney" suggestive of intussusception. Contrast CT abdomen confirmed ileo colic and colocolic intussusceptions which appeared as sausage shaped masses, bowel within bowel pattern.[fig:1.2]
A final diagnosis of small bowel obstruction/intussusception with anaemia was made. Surgeons opinion was sought and management plan charted out. As the patients haemoglobin was 6.0 gms% packed red blood cell transfusion and correction of anaemia followed by surgical intervention was planned.

With the informed and written consent of the patient’s attendants the patient underwent exploratory laparotomy. The intraoperative findings was ileocolic and colocolic intussusception. Careful manual reduction was done and a 2cms size polyp was noted as a lead point in the small bowel and resected with a section of bowel on either side followed by a 'clean sweep' procedure of intra operative enteroscopic polypectomy of polyps with size > 1cms was performed. [fig:3,4] Larger polyps were removed by enterotomy and followed by meticulous closure of the same and enterooristeromy of the resected small bowel. Post operatively the patient recovered well and was discharged on the 10th post op day.

Histopathological examination: [fig:5,6] B-1663-11 dated 19.08.11 reported - sections studied showed polypoidal lesions lined by small bowel mucosa/colonic, a core of arborising glands supported by broad bands of muscle fibres thicker in the centre and thinner towards the periphery the glands consist of columnar cells goblet cells, paneth cells and endocrine cells. Few glands are cystically dilated with mononuclear infiltrates. Numerous capillaries seen in the mucosa and submucosa. Features are of hamartomatous poly highly suggestive of Puetz Jeghers polyp.

The patient was followed up regularly for eight months, he had no recurrence of pain abdomen or bleeding per rectum and his nutritional status had improved considerably. However the patient was lost for follow up after that.

**Fig -1 : CT Scan image showing ileocolic/colocolic intussusception.**

**Fig-2: CT Scan showing ileo colic intucusseption with a soft tissue as a lead point.**

**Fig:3- operated specimen showing resected segment of small bowel with polyp.**

**Fig:4-operated specimen showing multiple polyps within the lumen.**
DISCUSSION:

Peutz-Jeghers syndrome was first described by Puetz in 1921 and Jeghers in 1944 and 1949[1]. This syndrome is characterised on physical examination by mucocutaneous pigmentation usually occurring during infancy and fading in late adolescence[1,2,4], as seen in our patient. The brownish pigmentation are seen on the vermilion border of lips(94%), buccal mucosa(66%), hands(74%), feet(62%)[5], periorbital, perianal and genital pigmentation noted in 95% of the cases caused due to pigment laden macrophages in the dermis [7]. Peutz-Jeghers occurs equally in male and female sex and is diagnosed during childhood or early adulthood.

It is an autosomal dominant disease considered rare with prevalence estimates ranging between 1:25,000-1:300,000 populations and in India 4,260 per 1,065,070,607 population.[3] Spontaneous mutation to STK 11 tumour suppressor gene on chromosome 19p13.3 have been shown to cause Peutz Jehgers syndrome.[2,3]

Peutz-Jeghers polyps are true hamartomas with a unique histological features such as branching frame work of connective tissue and muscle lined by intestinal epithelium rich in goblet cells. The polyps have elongated and convoluted glands with arbourising pattern and growth. These polyps occur numerously in small intestine (64%), stomach(49%), colon(64%), rectum(32%).[7] Polyps numbering between 1-20 per segment of gastrointestinal tract varying in size from 1-5 cms may be seen.[6] Such polyps are known to occur in extra intestinal sites such as renal pelvis, urinary bladder, lungs and nares, gall bladder[9,10]

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Endoscopic polypectomy, Colonoscopic polypectomy and 'Clean Sweep' procedures are an alternate modalities in reducing the repeated surgeries in patients with Pueetz Jeghers Syndrome.

Treatment of Peutz Jeghers syndrome should include a combination of endoscopy and laparotomy/laparoscopy.

REFERENCES: