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Case Report

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

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ABSTRACT

PeutzJeghers syndrome is a condition in which multiple hamartomatous polyps are present in the gastrointestinal tract in association with distinctive mucocutaneous pigmentations. 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 PuetzJeghers syndrome often present with history of pain abdomen due to small bowel obstruction / intusucception . 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 PeutzJeghers Syndrome with sub acute small bowel obstruction due to recurrent intusucceptions. He underwent laparotomy with reduction of intusucceptions and resection and anastomosis along with intraoperative enteroscopicpolypectomy. 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.

1. Introduction

PuetzJeghers syndrome is an autosomal dominant disorder characterised by presence of multiple gastrointestinal polyps and an increased risk for cancers.[6] The hamartomatous polyps are found most frequently in the small bowel, stomach and colon.The diagnosis of peutzjeghers is made in patients with at least two of the following clinical criteria: mucocutaneous melanin deposits, family history of PuetzJeghersSyndrome ,and small bowel polyposis.[4]

The PeutzJeghers polyp is a true hamartoma with unique histopathological characteristics such as frond like structure,appropriate epithelium for the area of gastrointestinal tract and smooth muscle proliferation.[7]

These polyps grow during the first decade of life and most patients become symptomatic between10-30years. The average age of peutzjeghers syndrome is 23years in men and 26years in women.The most frequent complication of peutzjeghers syndrome is intusucception occurring in 47% of patients and primarily in small bowel-95%.[7] We report a case of recurrent multiple intusucceptions as a sequelae of peutzjeghers syndrome who presented with chronic anaemia and pain abdomen.

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2. CASE ILLUSTRATION:

A 22 years old patient was brought to Medical out patientdepartment 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 peutzjeghers syndrome with sub acute small bowel obstruction secondary to intusucception 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. Ultrasonography showed a heterogenous mass in the epigastric region measuring 4x5.4x6cms giving an appearance of a “pseudo kidney”suggestive of intusucception. Contract CT abdomen confirmed ileo colic and colocolicintusucceptions which appeared as sausage shaped masses,bowel within bowel pattern.[fig: 1.2]

A final diagnosis of small bowel obstruction /intusucception 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 laporotomy. The intraoperative findings was ileocolic andcolocolicintusucception. 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 enteroscopicpolypectomy 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 enteroenterostomy 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-1663-11Dated 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 polyp highly suggestive of PuetzJeghers 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/ colocolicintusucception.

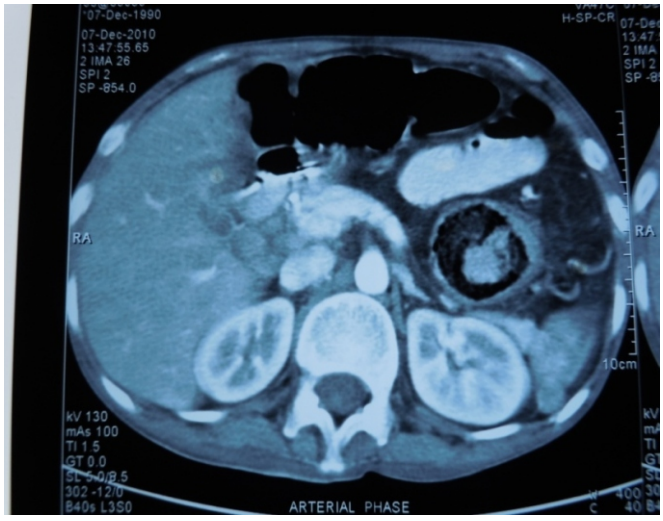


Fig-2: CT Scan showing ileo colic intucusseption with a soft tissue as a lead point.

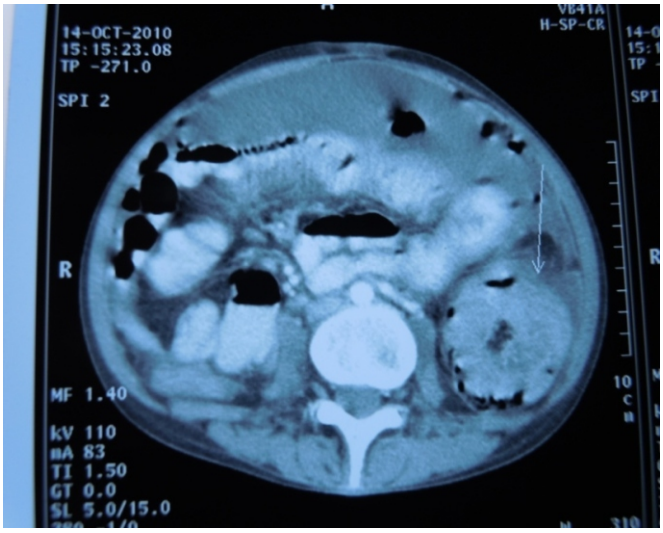


Fig:3- operated specimen showing resected segment of small bowel with polyp.

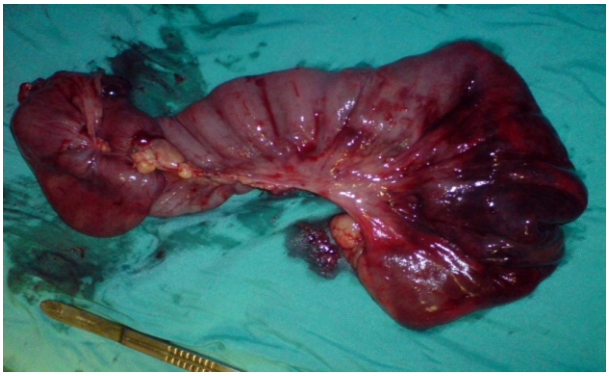


Fig:4-operated specimen showing multiple polyps within the lumen.

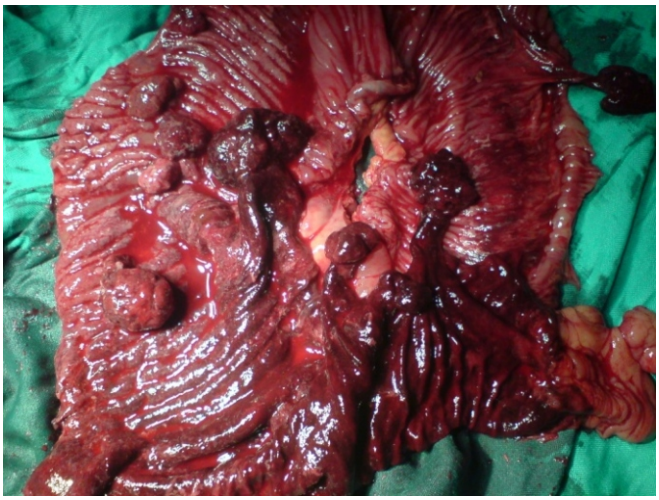


Fig: 5: Microphotograph showing arborisation arising from the muscularis mucosa in the polypoid structure (H&E x400)

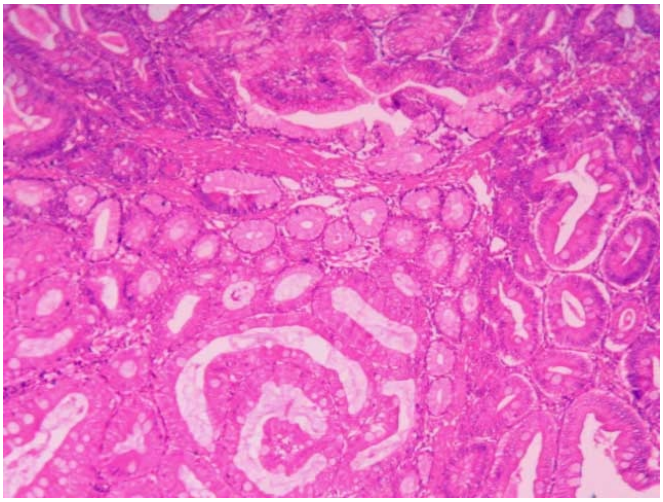
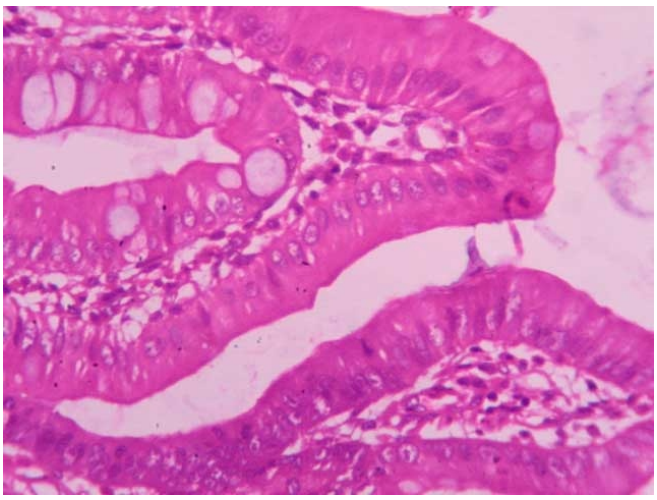


Fig:6: Microphotograph showing polypoid structure lined by simple columnar and goblet cells (H&E X400)



DISCUSSION:

PuetzJeghers 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 pigmentations are seen on the vermillion border oflips(94%),buccal mucosa(66%), hands(74%), feet(62%)[5], periorbital, perianal and genital pigmentationis noted in 95% of the cases caused due to pigment laden macrophages in the dermis [7]. Peutzjeghers 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 1:250,000-1:300,000 populationsand 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 Puetzjeghers syndrome.[2,3]

Puetzjeghers 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]

Puetzjeghers polyps grow during first decade of life and most become symptomatic between 10-30 years.[7] Average age at diagnosis for men is 23 years and 26 years for women. Our patient presented at 22 years. The modes of presenting are intestinal obstruction(43%), pain abdomen(23%), passing blood in stools(14%),and rectal mass/polyp(7%). In 13% of the patients it is diagnosed by melanin pigmentation. Our patient presented with all the features and in addition he was anaemic, this is due to the fact that the polyps ulcerate leading to acute blood loss or chronic anaemia.

Peutzjeghers syndrome patients usually present with intermittent pain abdomen caused by polyps. This feature was seen in our case from 1 year on and off, which can be explained by the fact that there must have been spontaneous reduction and it was a habitual/chronic intusucception.

Patients withPeutzjeghers syndrome are at risk of developing gastrointestinal cancers of 2%-3% diagnosed at a mean age of 40 years. They also have an increased risk for extraintestinal malignancies such as pancreatic,breast,ovarian and testicular cancers. Therefore it is mandatory to follow up regularly. The protocol should include GI endosopy, colonoscopy, small bowel contrast series at 12, 18 and24 years along with breast examination of female patients and testicular examination of males at regular intervals.

Various surgical and non surgical options are available now so also for diagnosis such as capsule endoscopy. Laparotomy is indicated in symptomatic patients with intestinal obstruction /intusucception. Some experts suggest that an attempt to clear the small bowel 'clean sweep' should be made during laporotomy with assisted intraoperative enteroscopy and polypectomy of all detected polyps and those > 1 cms.[6,11] We performed manual reduction of intusucceptions and resection of bowel was inevitable because of the sessile nature of the polyp as a lead point with ulceration and pre gangrenous changes. 'Clean sweep' procedure has the advantage of preventing recurrent intusucceptions and repeated surgical resections which can otherwise lead to short gut syndrome.[13]

CONCLUSION

PuetzJegherssyndrome caused by germline mutation of STK11 gene is an autosomal dominant condition characterised by small bowel hamartomatous polyposis and mucocutaneous melanin pigmentation.

Whenever surgery becomes inevitable care should be taken in adopting a proper approach and to preserve as much intestinal length as possible as these are patients who have a life long disease and may require repeated operations.

Regular follow up / surveillance is recommended right from the age of diagnosis because of the reported cancer risks.

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

Treatment ofpeutzjeghers syndrome should include a combination of endoscopy and laporotomy/laparoscopy.

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