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PEUTZ-JEGHERS SYNDROME WITH RECURRENT INTUSSUSCEPTION- OUR EXPERIENCE

Surgery

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ABSTRACT

Peutz-Jeghers' syndrome (PJS) is an autosomal dominant disease, which is characterized by mucocutaneous melanin pigmentation and multiple gastrointestinal polyps (hamartomatous). The gastrointestinal polyp backdrop may act as lead point for intussusception. Attempt at surgical intervention may need multiple resections leading to short bowel syndrome, adding to morbidity in these cases. We are herewith presenting a case of Puetz –Jeghers syndrome with recurrent intussusception.

KEYWORDS

Peutz-Jeghers syndrome, intussusception, melanosis, polyps

Introduction

Peutz-Jeghers syndrome (PJS) is an inherited autosomal dominant disorder characterized by multiple hamartomatous polyps in the gastrointestinal tract (<100) and melanotic mucocutaneous lesions. According to the literature available, Peutz reported the first case in 1921 and syndromic description was completed by Jegher in 1949 . Bruwer coined the eponym "Peutz-Jeghers" in 1954 . The incidence is 1 in 30,000 to 120,000 live births and there is no sex predilection noted . PJS description in literature was vast but its clinical exposure was very rare that too in Indian population. Here we are reporting a case of Puetz-Jeghers syndrome with multiple GI polyposis with recurrent intussusception.

Case report

A 20-year male presented to the emergency room (ER) with complaints of diffuse colicky abdominal pain, vomiting and constipation from 2 days. On arrival to triage his vitals were as follows- afebrile with pulse rate 114/min, blood pressure 100/70 mm of Hg and Spo2-99% on room air. He was triaged as ESI level 3 and evaluated as described below.

First detailed history and examination was carried out. There was no history of vomiting, hematemesis and melena. His previous medical records showed that around 7 years ago, he had similar complaints for which he underwent resection and anastomosis of bowel. Resected specimen was documented to contain multiple hamartomatous polyps of various sizes. No history of cancer-related deaths in the family.

On general examination, multiple melanotic pigmented spots were found over lips and buccal mucosa (figure 1)which according to the patient were present since birth. He was dehydrated on presentation and had severe pallor. On examination of abdomen, there was tenderness present with guarding and no rigidity was noted. Mild fullness was noted around umbilicus with an ill defined firm mass of around 5x5cm present on palpation. The mass was mobile from side to side and was not moving with respiration. No visible peristalsis was noted and it was resonant on percussion. There was no free fluid in abdomen and sluggish bowel sounds were found on auscultation. Per rectal examination was done and had multiple polypoidal growths in the rectum.

Clinically intestinal obstruction diagnosis was made and routine blood investigations were done along with imaging (X-Ray abdomen erect and ultrasound abdomen). Erect X-Ray abdomen showed multiple dilated bowel loops with a pattern of small bowel obstruction(multiple air fluid levels). Ultrasound showed 18cm long, bowel within bowel appearance with dilated proximal bowel loops. Maximum calibre of the dilated bowel was about 3.8cm. The classical ultrasound examination finding of "target sign" was observed in distal ileum (Ileo-ileal intussusception) (figure 2). Contrast-enhanced computerised tomography (CECT) abdomen done showed multiple polyps in small intestine, large intestine and rectum. The polyps were of variable sizes with the maximum being 5.5 cm found in the large bowel (figure 3).

Patient was managed conservatively with nasogastric tube decompression, nil per oral and correction of fluid and electrolyte disturbances. The in-hospital course was uneventful and he was discharged from hospital after 10 days. Before the discharge, he underwent colonoscopy which showed multiple polyps of various sizes in large intestine and rectum. Biopsy was taken and sent for histopathological examination which showed that the polyps were hamartomatous without evidence of any malignant changes. With hypermelanotic macules of the oral cavity and multiple hamartomatous polys throughout the intestine, we arrived at a diagnosis of Peutz-jeghers syndrome. Consequently, the patient and his family were referred for further genetic evaluation. Patient and patient attendants were also educated about future risks of malignancy and need for frequent follow-ups.

Fig 1 : hyperpigmented macules over lips and oral cavity

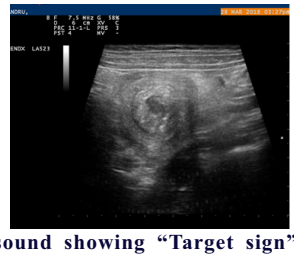

Fig 2 : Ultrasound showing "Target sign" suggestive of intussusception

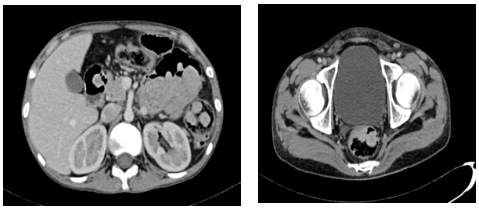

Fig 3 : CT Abdomen showing multiple polyposis of stomach, duodenum, colon and rectum

Discussion

PJS is a type of intestinal polyposis syndrome mainly characterized by the presence of melanotic pigmented macules over oral cavity mucosa and hamartomatous polyps of the gastrointestinal tract. It is basically an autosomal dominant disease with both the sporadic or familial pattern of inheritance. The familial pattern is the most common pattern comprising of 80% but sporadic pattern comprises 20%. Gene responsible for the familial pattern is serine-threonine kinase (STK) located on the short arm of chromosome 19 (19p33). This gene acts as a tumour suppressor gene and its mutation leads to uncontrolled growth patterns as polyposis and melanotic macules. Sporadic cases contain new onset mutation of STK11 gene with low penetration and doesn't contain any familial history.

Major criteria for the diagnosis of PJS consists of melanotic macules and hamartomatous polyps of GIT According to literature available, incidence rate is 1in 2,00,000. Mucocutaneous melanotic macules are present in most cases of PJS but its absence is also noted in some cases. They develop in infancy and childhood but gradually fade in adults. Rarely they may be found in adults . Most common location of these macules is mucosa of oral cavity but one may also notice them over lips, fingers, palms, soles, mucosa of the intestinal tract and perianal region. These macules are considered to be non-premalignant lesions. Coming to gastrointestinal (GI) polyps which are histologically hamartomatous in nature most common location for them is small intestine. But their presence can be expected from stomach to rectum. GI polyps starts occurring by 11-13years of age but by the age of 20 almost 50% of patients have symptomatic polyps. These patients often present with colicky abdominal pain due to recurrent intussusception caused by them. The reason behind this is polyps act like lead points for intussusception to happen. They may go for ulceration with chronic occult blood loss causing anemia. Another frequent presentation noticed is like small bowel obstruction. There are chances of extraintestinal polyposis expected in sites such as gallbladder, kidney, ureter, bronchus and nasal cavity.

Average size of these polyps vary between less than 1cm to more than 3cm. Histologically these polyps contain central widely arborizing smooth muscle covered by native intestinal mucosa with abnormal growth pattern. There is a tendency that these polyps turns into adenomas or carcinomas over time. So repeated colonoscopic examination for early identification of malignant transformation is necessary. Extraintestinal malignancies involving pancreas, breast, uterus, cervix, ovary, testis, lung and thyroid are fairly common in these patients.

Diagnosis of PJS can be confirmed by histological examination of hamartomatous polyps along with two of the following major clinical criteria- family history, mucocutaneous pigmented macules and GI polyposis. .

Imaging that is typical of PJS consists of identification of polyps in the stomach, small bowel and colon done by barium studies, ultrasound or CT non-invasively. Endoscopy also identifies polyps and also provides the chance of getting tissue for diagnosis.–

Treatment of polyps is an important aspect of management of PJS as it prevents both complications due to polyps and malignant transformation. Intraoperative endoscopy (IOE) and Double balloon endoscopy are the two modalities of treatment. IOE is more invasive and includes endoscopy of the entire bowel at the time of surgery followed by polypectomy. DBE allows visualizing almost entire bowel due to advanced ergonomics of equipment by preventing bowel kinking and further allows polyp resection. Screening for polyps can be performed either by enteroclysis or by capsule endoscopy. Capsule endoscopy gives the scope of examination of the entire bowel even well before obstructive symptoms started. This capsule endoscopy prevents short gut syndrome probability indirectly by preventing the risk of frequent bowel resection(6).\

Conclusion

Recurrent intussusception is the major morbidity of Puetz-jeghers syndrome due to its multiple polyposis of GI tract. Malignant transformation of polyps will be the additional burden. Close follow up of cases is necessary along with endoscopy at frequent intervals to detect early malignant transformation. This is the cornerstone of conservative management.

Informed Consent

Written informed consent was obtained from the patient for publication of his personal data.

Conflict of Interest

There is no conflict of interest.

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