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Case report

A rare case of multiple intussusceptions in Peutz–Jeghers syndrome

P.N. Sreeramulu and Vijay P. Agrawal

Department of General Surgery, Sri Devraj Urs Medical College, Tamaka, Kolar-563101, Karnataka, India.

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Corresponding author

Vijay P. Agrawal
Department of General Surgery,
Sri Devraj Urs Medical College,
Tamaka, Kolar-563101,
Karnataka, India.
Email: vijugunnu@gmail.com

Abstract

Peutz–Jeghers syndrome is a rare autosomal dominant genetic disorder characterized by the development of benign hamartomatous polyps in the gastrointestinal tract and hyperpigmented macules on the lips and oral mucosa. We present a rare case of Peutz-Jeghers syndrome with multiple intussusceptions. A 30 year old female was brought to the emergency room with colicky abdominal pain in umbilical area and lower abdomen, vomiting and blood in stools since 1 day. The lower lip and sole of feet showed hyperpigmented patches. On examination, bowel sounds were increased and rectal examination revealed black tarry stool. Exploratory laprotomy done revealed jejuno-ileal and ileo-cecal intussusceptions for which intestinal resection and anastomosis was done. There were multiple polyps in the resected ileal segment.

Key words: Peutz-Jeghers syndrome, multiple intussusception

Peutz–Jeghers syndrome is a rare autosomal dominant genetic disorder characterized by the development of benign hamartomatous polyps in the gastrointestinal tract and hyperpigmented macules on the lips and oral mucosa. The risks associated with this syndrome include a strong tendency of developing cancer in multiple sites. While the hamartomatous polyps themselves only have a small malignant potential, patients with this syndrome are at greater risk of developing carcinomas of the pancreas, liver, lungs, breast, ovaries, uterus, testicles and other organs. Often the first presentation is as a bowel obstruction from an intussusception which is a common cause of mortality. We present a rare case of Peutz-Jeghers syndrome who presented with multiple intussusceptions.

Case report

A 30 year old female was brought to the emergency room with colicky abdominal pain in umbilical area and lower abdomen, vomiting and blood in stools since 1 day. There was no history of fever and altered bowel habit. She was previously healthy and not on any medications. She had no family history of gastrointestinal disorders.

On examination, her vitals were stable. The lower lip and sole of feet showed hyperpigmented patches (Fig 1 and 2). There was tenderness and guarding in umbilical and lower abdominal area. Hernial orifices were normal. However, bowel sounds were increased. Rectal examination revealed black tarry stool. The other systemic examination was normal.

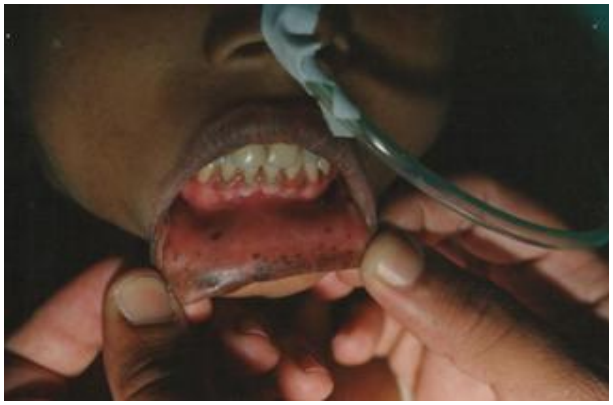

Fig 1. Lower lip showing hyperpigmented patches

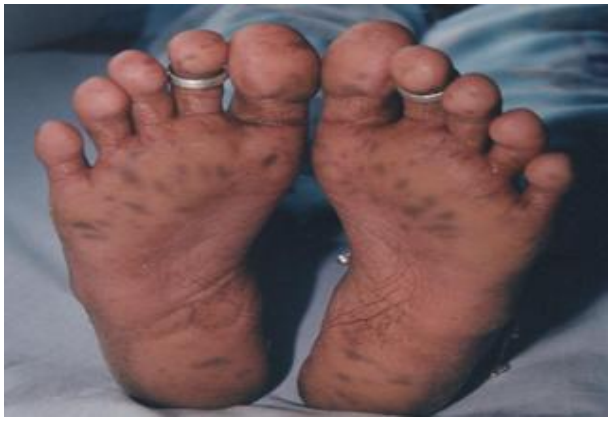

Fig 2. Sole of feet showing hyperpigmented patches

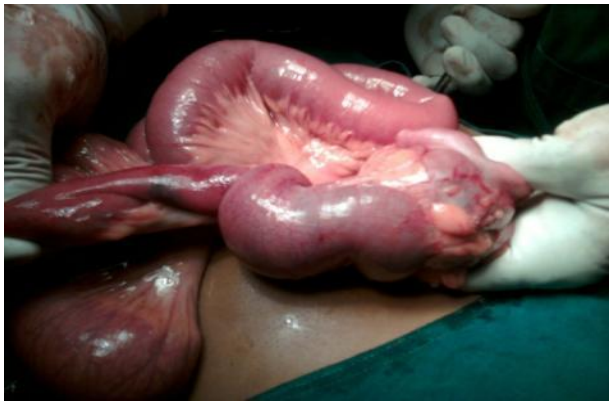

Fig 3. Jejuno-ileal intussusception

Her blood investigations were almost normal except for moderate anemia (Hemoglobin-7.5 gm/dl). A plain x-ray of the abdomen did not give much information, however abdominal ultrasonography showed features of multiple intussusceptions involving umbilical, right iliac fossa and left lumbar region.

Patient underwent exploratory laprotomy revealing jejuno-ileal and ileo-cecal intussusceptions (Fig 3 and 4). Resection of jejuno-ileal and ileo-cecal intussuscepted segment with jejuno-ileal and ileo-cecal anastomosis was done.

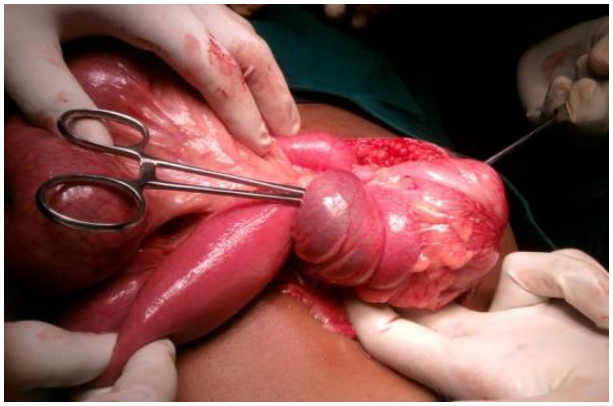

Fig 4. Ileo-cecal intussusception

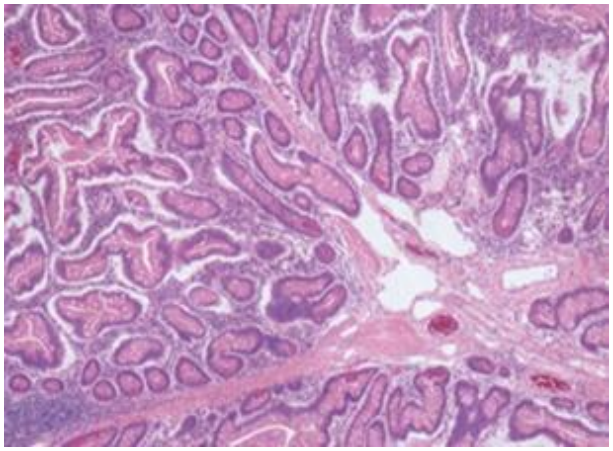

Fig 5. Focal epithelial hyperplasia, lamina propria smooth-muscle proliferation and arborizing architecture (Hemotoxylin & Eosin stain)

There were multiple polyps on the ileal resection margins. Histopathological examination revealed features of multiple Peutz-Jeghers polyps with no gangrenous and malignant changes (Fig 5). Patient's postoperative course was uneventful and is doing well with 6 months of follow up.

Discussion

Peutz-Jeghers syndrome was described in 1921 by Jan Peutz, a Dutch physician who noted a relationship between the intestinal polyps and the mucocutaneous macules in a Dutch family. In 1949, Harold Jeghers an American physician was credited with the definitive descriptive reports of the syndrome. It is a rare autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition[1]. Peutz–Jeghers syndrome has an incidence of approximately 1 in 12-30,000 births[2].

The cause of Peutz-Jeghers syndrome (PJS) in most cases (66-94%) appears to be a germline mutation of the STK11/LKB1 (serine/threonine ki-

nase 11) tumor suppressor gene, located on band 19p13.3[3].

The diagnostic criteria for the syndrome are as follows:[4]

a) Three or more histologically confirmed Peutz-Jegher's polyps or
b) Any number of Peutz-Jegher's polyps with a family history or
c) Characteristic prominent mucocutaneous pigmentation with a family history or
d) Any number of Peutz-Jegher's polyps with characteristic prominent mucocutaneous pigmentation.

During the first 3 decades of life, anemia, rectal bleeding, abdominal pain, obstruction, and/or intussusception are common complications[5,6]. The occurrence of adult intussusception is rare[7-9]. In adult, intussusception most commonly involves small intestine whereas in children, intussusception is common and most commonly involves large intestine[10].

In a series of 222 patients with Peutz-Jeghers syndrome (PJS), Utsunomiya et al noted the following distribution of presenting gastrointestinal symptoms:[11]

- Obstruction - 42.8% of patients
- Abdominal pain caused by infarction - 23% of patients
- Rectal bleeding caused by ulceration - 13.5% of patients
- Extrusion of polyp - 7% of patients

Polyps in the Utsunomiya study occurred as follows:

- Small intestine - 64% of patients
- Colon - 63.2% of patients
- Stomach - 48.6% of patients
- Rectum - 32% of patients

The incidence of polyps within the small intestine is greatest in the jejunum and progressively decreases in the ileum and duodenum[6].

Other rare reported complications include gastrointestinal obstruction and vomiting secondary to gastric polyps. Extraintestinal polyps are also reported although they are rare; they include nasal polyps, gall bladder polyps, ureteric polyps and respiratory tract polyps[12].

Cancer develops in the gastrointestinal tract of patients with PJS with a higher frequency than it does in the general population. They have a 15-fold increased risk of developing intestinal cancer compared with that of the general population[2]. The major sites of cancer occurrence are small intestine, stomach, pancreas, colon, esophagus, ovary, lung, uterus, breast and cervix[13].

Surgical treatment of extraintestinal cancers detected by surveillance and diagnosis is required. Laparotomy and resection may be necessary, as indicated, for small intestinal intussusception, obstruction or persistent intestinal bleeding.

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Conflict of interest: None

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