

A Rare Case of PeutzJegher Syndrome Presenting with a Solitary Polyp in the Jejunum – A Case Report

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Abstract: Background: Intussusception is a common abdominal emergency requiring surgical intervention in newborns and infants. They commonly present in babies less than 2 years of age. A vast majority of intussusceptions are idiopathic. They rarely present in older age groups and in adults. Certain autosomal disorders may be linked to intussusceptions. Peutz Jegher Syndrome is a rare autosomal dominant disorder resulting due to mutation in the SPK 11 gene located in chromosome 19q13.3. The disease is characterized by multiple hamartomatous polyposis, jejuna intussusceptions and hyperpigmented melanotic spots in the mucocutaneous regions like mouth, etc. Case report: We present a case of 12 year old girl with chronic stomach pain and occasional vomiting. General examination showed presence of hyperpigmented spots in the soles, cheeks, lips and mouth. CECT abdomen showed presence of jejunal intussusception. The patient was taken up for laparoscopy and laparotomy was performed after reducing the intussusceptions. A solitary sessile polyp was removed and histopathological examination confirmed the presence of hamartomatous polyp. There is an increased susceptibility of cancers of gastrointestinal system and several other organs like breast, ovaries, etc. Conclusion: Further research in this area may be carried out to explore the risk factors and genetic mechanisms which may help in early detection and prevention of such rare syndromes.

Keywords: Hamartomatous Polyp, Jejunal Intussusception, Laprotomy, Peutz Jegher Syndrome

1. Introduction

Intussusception is a surgical emergency occurring as a result of invagination of bowel into an immediately adjacent section of the bowel. It is characterized by telescoping of a part of intestine into itself. [1] Intussusception is a common cause of abdominal pain in the pediatric age group. It is however, the most common surgical emergency among the children all over the world. High incidence rates are seen in infants aged 4-10 months. [2] The diagnosis is often clinical, although radiological procedures like CT and MRI of the abdomen might help. [3] There are several risk factors attributed in children namely infections, cystic fibrosis and intestinal polyps. Some of the other risk factors include anatomical abnormalities, Meckel's diverticulum, altered mobility, duplication, appendicitis and hyperplasia of Peyer's

patches.

Intussusception often presents at five months of life, peaks at around nine months with the lowest incidence rates reported at 18 months. [4] The most common end point pathology in intussusceptions is intestinal obstruction. Peristaltic movements play a major role in the pathophysiology of intussusception. The peristaltic action pulls the proximal segment into the distal segment. There is often an anatomical lead point noted in 10% of the intussusceptions. Intussusception presents with vomiting, abdominal pain and rectal bleeding. There are several maneuvers to reduce the intussusception. However, in most cases, surgical correction is often warranted.

In rare situations, intussusception is caused by certain genetic and autoimmune disorders. Peutz Jegher Syndrome is an autosomal dominant disorder characterized by various presentations like mucocutaneous hyperpigmentation and

hamartomatous polyp in the gastrointestinal tract. They are also associated with intussusceptions. [5] In this case report we present a case of Peutz Jegher syndrome which presented as jejuna intussusception in a 12 year old girl.

2. Case Report

A 12 years old female, presented with history of chronic, recurrent pain abdomen on and off. There were also complaints of vomiting, which was bilious in nature, on and off for 10 days. The patient did not have any other abdominal symptoms. On examination, the general condition was good, the patient was neither anaemic nor icteric. There were presence of a few hyperpigmented spots in the soles, cheeks, buccal mucosa and lips (figure 3). Systemic examination of cardiovascular and respiratory system did not reveal any abnormality. Abdomen examination revealed a vague mass in the left hypochondrium. There was no tenderness. The mass was moving with respiration and firm in consistency. The extrinsic mobility was however restricted. Rectal examination did not reveal any abnormality.

Ultrasonogram of the abdomen revealed the presence of a mixed echogenic mass lesion in the left hypochondrium. There was evidence of telescoping of bowel loops (bowel within the bowel) in the left lumbar and hypochondrial region. A small hypoechoic rounded lesion of size 3.5x2.5 cms adjacent to the loop was found with internal vascularity. A provisional diagnosis of colocolic intussusception was made. Colonoscopy was done and did not reveal any abnormality up to the hepatic flexure. Contrast Enhanced CT showed Jejunojejunal intussusceptions with pseudo kidney sign (figure 1). Proximal stomach and duodenum were dilated. The distal passage of the contrast was visible.

The patient was taken up for surgical exploration. Under general anaesthesia, with 10mm telescope, two 5mm side ports, laproscopy was done. Jejunonjejunal intussusception was seen and was reduced. However, a mass was felt at 7-8 cm from DJ flexure. Therefore minilaprotomy was planned. Abdomen was opened through the umbilical port. A sessile polyp was noted and was resected and sent for histopathological examination (figure 2). End to end anastomosis was done. The post operative period was uneventful.

Histopathological examination showed hamartomatous mucosal polyp with characteristic central core of bracing smooth muscle associated with mucosa native of colonic origin. Polyp shows prominent arborizing smooth muscle bundle.

Considering the jejunal intussusceptions, hamartomatous polyp and hyperpigmented spots, a diagnosis of Peutz Jegher Syndrome was made.

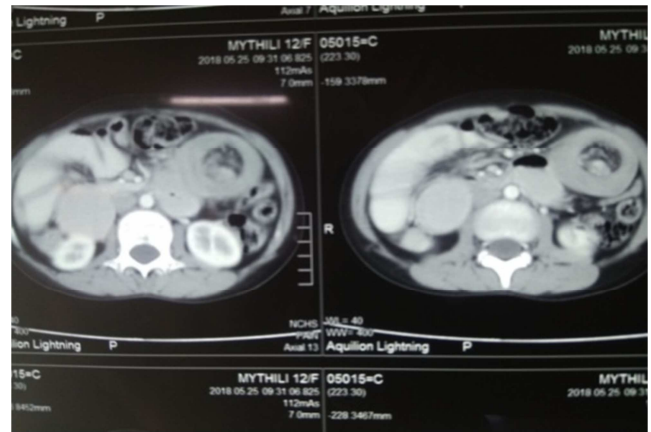


Figure 1. CECT abdomen showing jejuna intussusceptions.



Figure 2. Hamartomatous polyp.

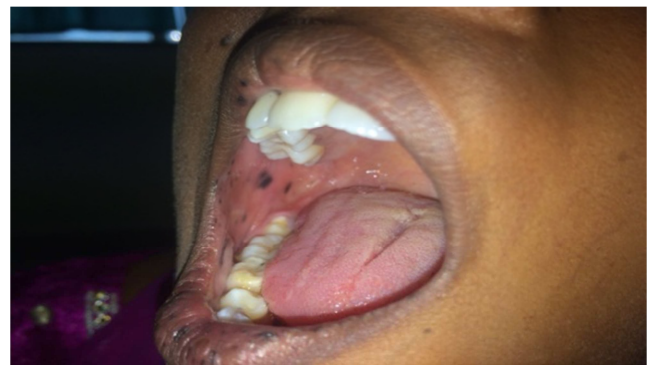


Figure 3. Mucocutaneous hyperpigmented spots.

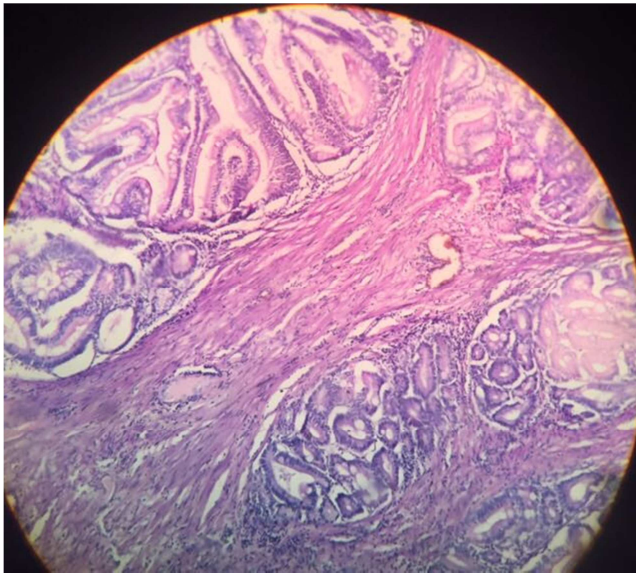


Figure 4. Hamartomatous mucosal polyp with characteristic central core of branching smooth muscle associated with mucosa native of colonic origin. Polyp shows prominent arborizing smooth muscle bundle.

3. Discussion

PeutzJegher syndrome (PJS) is an autosomal dominant disorder first reported by Peutz in 1921. [6] It was Brunner and his associates who described the clinical features of this syndrome. The syndrome is characterized by presence of hamartomatous polyps in the gastrointestinal tract and pigmented mucocutaneous lesions. It belongs to the familial hamartomatous polyposis syndromes which are associated with significant mortality and predisposition to certain malignancies. The hamartomatous polyposis syndromes are heterogenous disorders and represent <1% of the inherited gastrointestinal cancer predisposition syndromes. [7, 8]

PJS is a rare syndrome occurring in 1 in 100,000 people. [9] There are very few studies which have documented the prevalence of PJS. Most of the known cases are presented as case reports. Being an autosomal dominant disorder, the genetic involvement in this disease is said to be a result of mutation in the STK11 (serine/threonine-protein kinase or LKB1) gene located on the chromosome 19p13.3. This gene encodes for the role of second messenger in signal transduction. It modulates cellular proliferation, controls cell polarity and has a role in responding to low energy levels. [10] The STK11 gene is also involved in inhibition of AMP – activated protein kinase and signals to inhibit the mammalian target of rapamycin (mTOR). Overall STK11 codes for a protein product which is responsible for growth inhibition. Therefore, genetic mutation in this gene will result in loss of heterozygosity at a tumor suppressor gene locus and thereby increase the susceptibility for cancers. [11]

The clinical features of PJS are characterized by melanotic spots, which are mucocutaneous lesions that are brown to black in colour. These spots are linear and irregular. They are usually small, measuring <5mm, flat, hairless and do not coalesce. Microscopically the pigments are seen in basal

layer of epidermis. The common areas include mouth, hands, feet, nasal mucosa, conjunctiva and rectum. The spots are usually present since birth. The lesions over lips and skin fade at puberty while those over buccal mucosa remain. Partial intestinal obstruction from transient intussusception and crampy abdominal pain remains the cardinal feature of PJS.

The hamartomatous polyps which occur in PJS are often multiple. Our study was a rare case where there was only a solitary, sessile polyp. Hamartomatous polyps are composed of normal cellular elements but have a distorted architecture. [8] Polyps are most frequently seen in small intestine, may also occur in nasal cavity, esophagus, stomach, colon, rectum, urinary bladder, bronchus. They are usually multiple and widely scattered. The size of the polyps ranges from few millimeters to several centimeters. The risk of developing colon cancer in the presence of hamartomatous polyps is 38%. Several genetic studies propose that these polyps are clonal premalignant lesions responsible for gastric carcinoma. [12]

The management of PJS requires recurrent laparotomies for gastric obstruction, resection of the polyps and for recurrent intussusceptions. Resection and anastomosis may be required for unreducible intussusceptions. Enterotomies may be done to remove the polyps. The patients are often anaemic due to malaena and chronic blood loss per rectum. Majority of the patients are susceptible to various cancers namely breast, pancreas, stomach, ovaries, lung and small bowel.

4. Conclusion

PeutzJegher Syndrome is a rare autosomal dominant disorder occurring in the childhood as a result of mutations in the SPK 11 gene. The disease, being characterized by hyperpigmented spots, recurrent intussusceptions and polyps is associated with higher rates of risk for gastrointestinal carcinoma and cancers of other organs like breast, ovaries, etc. There is a growing need to periodically screen these individuals, at least once a year for these malignancies. Being a genetically mediated disease, there is a dearth for research to be done to explore the screening modalities for detecting these polyps during the intrauterine period. It is also essential to evaluate for the risk factors and hazardous exposures, in order to prevent the incidence of such rare diseases.

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