ISSN 1996-3424

Asian Journal of Dermatology

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OPEN ACCESS Asian Journal of Dermatology

ISSN 1996-3424
DOI: 10.3923/ajd.2016.20.24

Case Report
Peutz-jeghers Syndrome in a Child Presenting with Acute Abdomen: A Case Report

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Abstract
A 13 year old female patient presented to paediatric OPD with complaints of pain abdomen and blood in stools. She was referred to Dermatology OPD for muco cutaneous hyperpigmented lesions on her palms, soles, oral mucosa and also on the knuckles. Keeping in hindsight the potential probability of puetz-jeghers syndrome (PJS), the patient was referred to surgery and investigations were carried out. After undergoing various investigations the diagnosis was confirmed. The patient underwent GI surgery for removal of the polyps.

Key words: Peutz-jeghers syndrome, pain abdomen, hemartomatous polyp

Received: November 08, 2015 Accepted: December 01, 2015 Published: December 15, 2015

Citation: Sundeep Chowdhry, Dipak D. Umrigar and Neha Yadav, 2016. Peutz-jeghers syndrome in a child presenting with acute abdomen: A case report. Asian J. Dermatol., 8: 20-24.

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Competing Interest: The authors have declared that no competing interest exists.

Data Availability: All relevant data are within the paper and its supporting information files.

INTRODUCTION

Peutz-Jeghers syndrome is a rare autosomal dominant disorder characterized by hamartomatous polyps and characteristic mucocutaneous pigmentation. The hamartomatous polyps of Peutz-Jeghers syndrome can cause intestinal occlusion and show preponderance in involvement of small intestine. Intussusception is seen frequently in children, but rarely in adults. Polyps are found throughout the gastrointestinal tract but most are confined to the small bowel (60-90%) and the colon (50-64%). These polyps may also be found at extra-intestinal sites such as the gallbladder, bronchi, bladder and ureter[1]. It can present as acute abdomen with other GI complaints. A strong clinical suspicion with clinically visible dermatological pigmentary changes and GI symptoms should prompt the physician to investigate for PJ syndrome.

The purpose of reporting this case is to remind clinicians that perioral or hand pigmentation should not be taken lightly in colorectal cases. The PJS also assumes more importance because of higher risk of intestinal and extraintestinal cancers[2].

Case presentation: We present a case of a 13 year old female child who came to paediatric OPD with complaints of pain abdomen and blood in stools since past 2 days. The patient had pigmentation on her lips, palms and soles. She gave history of multiple flat, dark coloured lesions over lips, oral cavity, face, palms and soles since past 7 years (Fig. 1 and 2). She also gave history of multiple dark coloured elevated skin lesions over both elbows (Fig. 3) and darkening of skin over both hands since last 3 months.

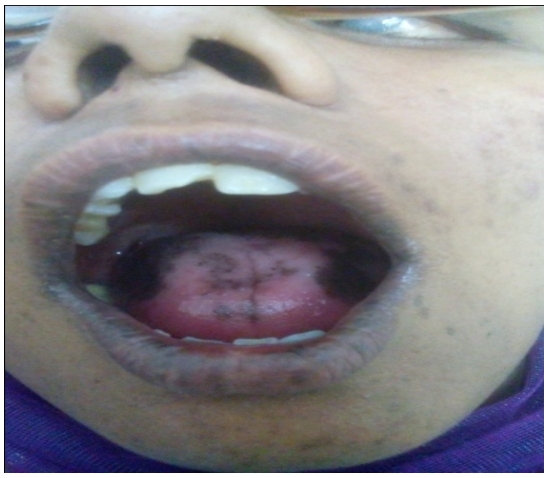

Fig. 1: Multiple discrete to coalescent hyperpigmented macules present over lips tongue and buccal mucosa

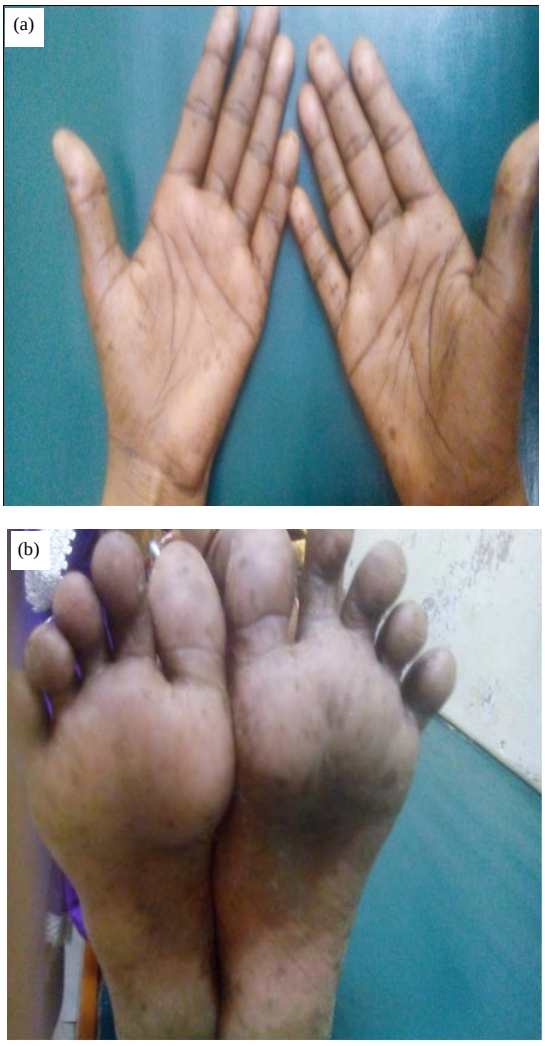

Fig. 2(a-b): Multiple hyperpigmented macules present over (a) Palms and (b) Soles

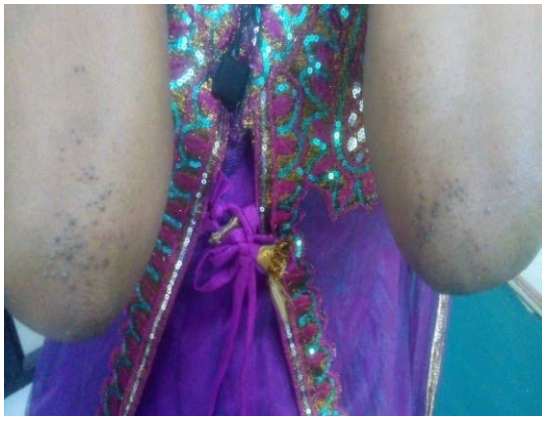

Fig. 3: Multiple hyperkeratotic skin coloured to hyperpigmented papule present over both elbows

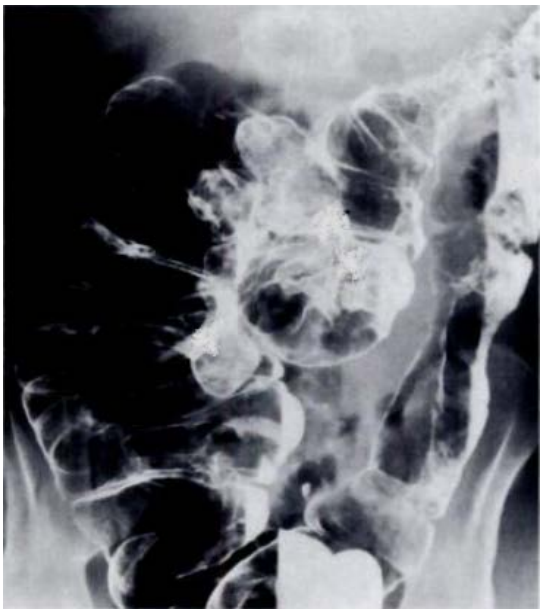

Fig. 4: Barium follow through study showed multiple intraluminal filling defects in the small bowel suggestive of multiple polypoid lesions in the small intestine

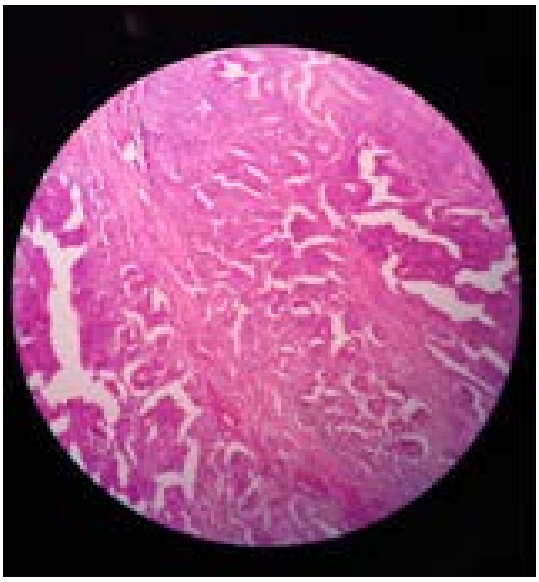

Fig. 5: HPE polyp revealed core of smooth muscle around which the intestinal mucosa is thrown in to folds giving tree like appearance

The patient also informed that similar skin lesions were present in her younger sister as well since past many years and her younger brother was normal and had no such lesions.

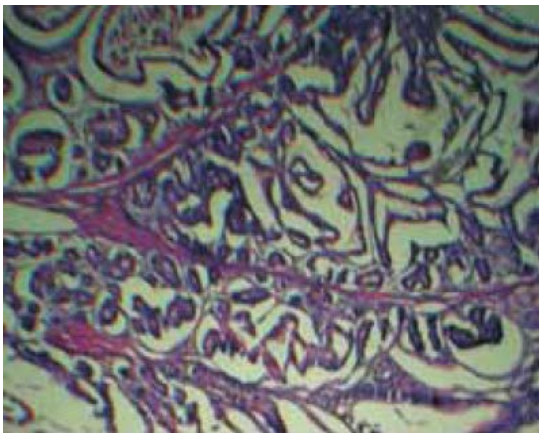

Fig. 6: HPE of Hamartomatous polyp demonstrating the arborizing pattern of smooth muscle proliferation upon resting the small intestinal mucosa

On examination, multiple 5-15 mm discrete to coalescent hyperpigmented macules were present over tongue and buccal mucosa. Also, multiple 3-7 mm sized hyperpigmented macules were present over lips, perioral area, palms and soles. Discrete hyperkeratotic skin coloured to hyperpigmented papules were seen over both elbows and both forearms. Hyperpigmentation was also present over knuckles and dorsal aspect of interphalangeal joints of hands.

The patient was referred to department of surgery for further investigations and evaluation.

Peripheral smear examination revealed microcytic hypochromic anaemia whereas the RBCs with hypersegmented neutrophils suggestive of dimorphic anemia. Serum vitamin-B12 level was 154 pg mL^{-1} (normal range 200-900 pg mL^{-1}), whereas, remaining routine biochemical parameters were within normal limits. Stool for occult blood test was positive. Abdominal USG findings were consistent with intussusception in small intestine. Barium follow through study showed multiple intraluminal filling defects in the small bowel suggestive of multiple polypoid lesions in the small intestine (Fig. 4).

The patient underwent an exploratory laparotomy and small bowel segmental resection with end to end anastomosis was performed.

The specimen was sent for histopathological examination. Microscopic examination of polyp revealed core of smooth muscle around which the intestinal mucosa is thrown in to folds giving tree like appearance (Fig. 5). Hamartomatous polyp demonstrating the arborizing pattern of smooth muscle proliferation upon which rests the small intestinal mucosa (Fig. 6).

DISCUSSION

The relationship of mucocutaneous pigmentation and intestinal polyposis was first reported in 1921 by dutch pysician Peutz[3] who studied a dutch family over three generations. Peutz noticed that seven of them had intestinal polyposis and four had nasal polyposis, besides the peculiar mucocutaneous pigmentation. After an initial report of two patients by Jeghers[4] the definitive clinical description of PJS was written by Jeghers *et al.*[5]. The authors also recognized that it was inherited as a simple Mendelian dominant trait. The PJS is a complex hereditary polyposis condition characterized by autosomal dominant inheritance, hamartomatous polyps of the gastrointestinal tract andcharacteristic mucocutaneous pigmentation. It is inherited in an autosomal dominant manner and is caused by a germline mutation in the STK11 (LKB1) gene which is located at 19 p13.3[6]. The incidence has been estimated as 1:1,20,000 births. This gene has been reported in 80% of patients with PJS. Up to 25% of recorded cases of PJS do not have family history. Those sporadic cases probably arise due to new mutation of STK11 gene or low penetration[7].

Fewer than 5% of patients with PJS lack the abnormal mucocutaneous melanotic pigmentation and fewer than 5% of patients with the pigmentation have no PJ polyps[8].

A clinical diagnosis of PJS may be made when any one of the following conditions is present in a single individual: Two or more histologically confirmed PJ polyps; any number of PJ polyps detected in one individual who has a family history of PJS in a close relative; characteristic mucocutaneous pigmentation in an individual who has a family history of PJS in a close relative; any number of PJ polyps in an individual who also has characteristic mucocutaneous pigmentation[1].

Gastrointestinal polyps in PJS are hamartoma. Hamartomatous polyps are composed of the normal cellular elements of the gastrointestinal tract, but have a markedly distorted architecture. The most common location for polyps is the small bowel (64%), although involvement of the colon (53%), stomach (49%) and rectum (32%) is also described[9].

Intussusception occurs when one loop of bowel (intussusceptum) telescopes into an adjacent segment (intussuscipiens). This clinical presentation has been observed in 47-69% of adult patients with PJS and most of them were due to polyps located in the small intestine[10]. The majority of PJS intussusceptions reported in the literature are ileal or jejunal[11-14]. Colo-colonic intussusception is reported in only a few cases[15]. The duodenum is a particularly uncommon site for intussusception, since it lies in a fixed retroperitoneal position.

The diagnosis of intussusception caused by PJS should be based on a history of PJS and physical examination. Abdominal distention and local tenderness are the most frequent findings. Abdominal symptoms tend to occur early in life, with more than 50% symptomatic patients before the age of twenty[16].

Proper abdominal examination may show an abdominal mass although it has been noted in only 12.5% of cases[12]. Abdominal CT has been reported to be the most useful imaging modality. The CT may help to delineate the precise location of polyps-differentiating betweenlead point and non-lead point intussusception which is important in determining the appropriate treatment and has the potential to reduce the prevalence of unnecessary surgical intervention (9). Endoscopy has a distinct role in the diagnosis and treatment of intussusception. Endoscopic polypectomy and double-balloon enteroscopy are therapeutic options even in patients with a history of extensive abdominal surgery. By the age of ten years, 30% of patients with PJS already required a laparotomy[17].

A rational therapeutic strategy for Peutz-Jeghers syndrome comprises endoscopic removal of any polyp larger than 5 mm. On the other hand, surgical treatment is indicated for removal of any small bowel polyps that are symptomatic or larger than 1.5 cm[16].

Double-balloon enteroscopy may decrease the need for laparotomy in patients with PJS[18,19]. Furthermore, the use of this approach can lead to a healthier life and longer life expectancy by avoiding short bowel syndrome in the case of multiple intussusception. However, this technique has limitations for large solitary polyps and needs expertise.

Though endoscopic removal is the ideal method of treating a pedunculated polyp; however, when this is not possible, laparoscopy can be a safe and effective alternative for reduction of the intussusception and bowel resection[20,21].

After diagnosis has been established, some authors recommend the following for patients with Peutz-Jeghers syndrome: Annual hemoglobin examination, annual ultrasonography of the pancreas, pelvis (women) and testes, upper and lower endoscopy per 2-years, small bowel X-ray every 2 years, mammography at the ages of 25, 30, 35, 38, thereafter per 2 years until the age of 50, then annually and cervical smear every 3 years. The interval between pancreatic ultrasonography and small bowel X-ray may be extended after several examinations yield negative results[16].

CONCLUSION

Intussusception caused by Peutz-Jeghers syndrome is a rare diagnosis and the asymptomatic skin lesions are often overlooked as being insignificant. It is important to carry out all relevant investigations, biochemical and radiological both, in a child having bowel complaints with hyperpigmented lesions on skin and mucosa as intestinal polyps or hamartomas are almost present in the small intestine which may lead to intussusception and obstruction warranting early and prompt surgical intervention.

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