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Case Report

Peutz-Jeghers syndrome: a case report

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ABSTRACT

Peutz-Jeghers Syndrome (PJS) is a rare, autosomal dominant disorder responsible for mucocutaneous pigmentation and gastrointestinal hamartomatous polyps. We present a case of Peutz-Jeghers syndrome in a 12 year old male child who presented with abdominal pain, vomiting and malena. The patient had pigmented lesions on the malar area, nose, lower lip and buccal mucosa. The imaging studies revealed multiple polyps in small intestine.

Keywords: Peutz-Jeghers syndrome, Child, Intestinal polyps

INTRODUCTION

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition. Patients with Peutz-Jeghers syndrome have a 15-fold increased risk of developing intestinal cancer compared with the general population.[1-9] The mutation is localized to chromosome 19p13.3.[10] An overall recommendation for PJS patients includes not only gastrointestinal multiple polyps resolution, but also regular lifelong cancer screening. Early detection and proper surveillance are vital to minimize the risk of gastrointestinal carcinoma.

CASE REPORT

A 12 year old boy presented to our department with complaints of multiple small hyperpigmented lesions on face and oral cavity since one year. He also complained of abdominal pain, vomiting and blood stained stools of two months duration. As stated by his father, the lesions initially started as asymptomatic small brown pigmented spots on the cheeks and nose and gradually involved the whole face within a period of 2 months (Figure 1). Later, they became darker and increased in size and eventually progressed to involve the lower lip and buccal mucosa (Figure 2). During this phase, patient also experienced intermittent abdominal pain since 2 months, which was diffuse and caused discomfort. He had few episodes of non-projectile vomiting and blood stained stools during the same period. There was no history of altered bowel habits, fever, anorexia, weight loss or any other significant systemic complaints. Family history was not contributory. On examination, there were well demarcated, diffuse, 2-3 mm dark brown macules present over entire face, densely distributed on malar area, lower lip & buccal mucosa. Other areas like trunk, extremities, back, external genitalia, gums and palate were spared. Scalp, hair, nails were normal. There was presence of significant pallor. On systemic examination, abdominal tenderness was present. Routine investigations revealed anemia (Hb-9 gm/dl) and stool was positive for occult blood. LFT, RFT, urine examination were within normal limits. USG & barium studies revealed small polyps in stomach with filling defect in jejunum & multiple small pedunculated polyps throughout the colon. USG pelvis

was normal. Endoscopy revealed 10-15 polyps in stomach, jejunum and colon (5-15 mm in diameter). Biopsy showed hyperplastic, dilated mucous glands and centrally radiating smooth muscle bundles with necrotic surface epithelium without any signs of malignancy (Figure 3).

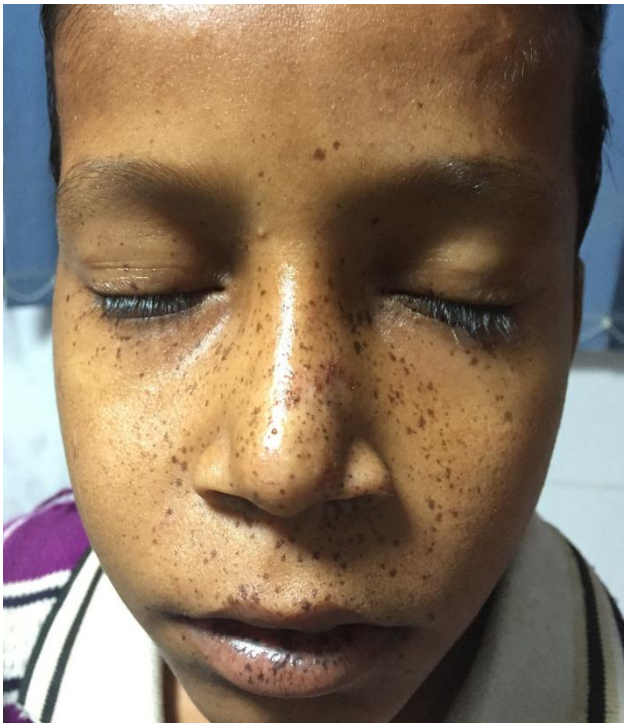

Figure 1: Multiple lentigens over the face.

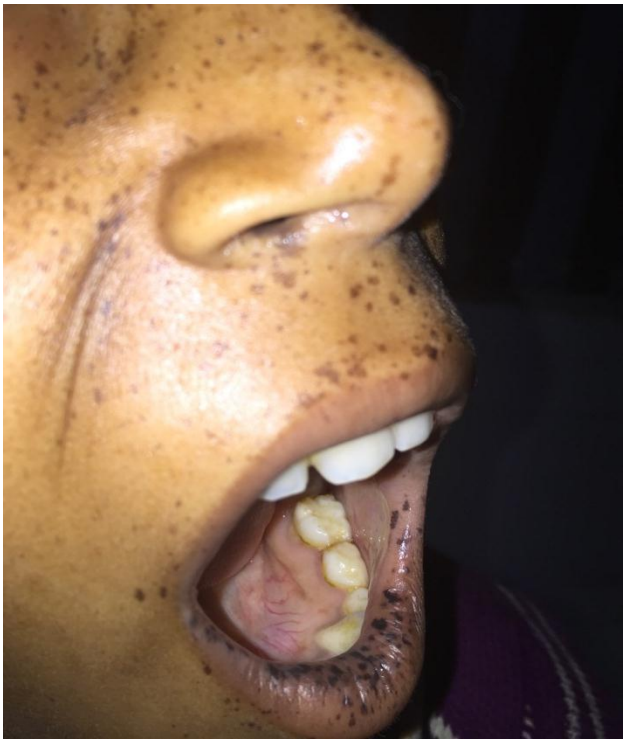

Figure 2: Lentigens on lower lip and buccal mucosa.

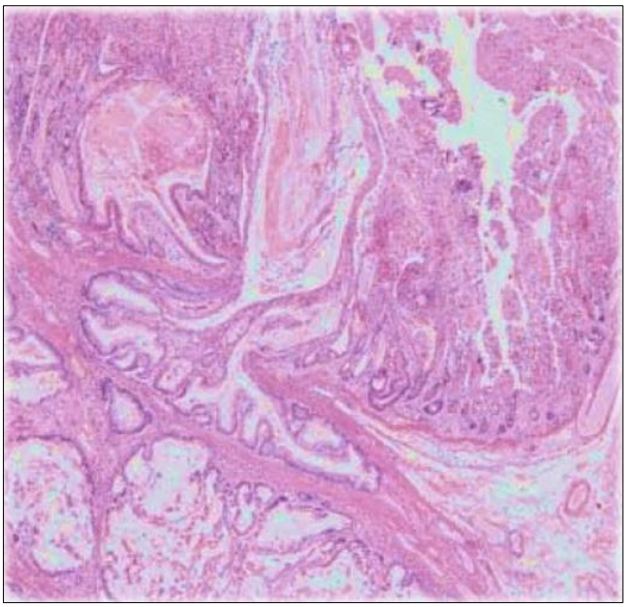

Figure 3: Hyperplastic, dilated mucous glands and central radiating smooth muscle bundles. Surface epithelium undergoes hemorrhagic necrosis due to intussusception.

DISCUSSION

The primary description of PJS was published by Peutz in 1921 in one Dutch family (the Harrisburg family) as a gastrointestinal familial polyposis with pigmentations.[11] Jeghers specified the description in 10 cases from different families in his work in 1949 and defined the relations between pigmented lesions, gastrointestinal polyposis and increased risk of carcinoma; approximately half of his patients suffered from gastrointestinal malignancy.[12] The Peutz-Jeghers syndrome consists of two major components: hamartomatous polyposis of the gastrointestinal tract and mucocutaneous pigmentation.[13,14] The incidence of PJS is reported to be 1 in 150000 to 200000 individuals.[15] The hyperpigmented lesions contain melanotic deposits and commonly manifest in infancy and childhood. Pigmented lesions can fade during puberty and adulthood.[13] The median time to first presentation with polyps is about 11-13 years of age and approximately 50% will have experienced symptoms by the age of 20 years.[14] Peutz-Jeghers polyps can also ulcerate, leading to acute blood loss or chronic anemia. Although Peutz-Jeghers polyps are most commonly found in the gastrointestinal system, they can also occur in extra-intestinal sites such as kidney, ureter, gall-bladder, bronchial tree and nasal passages. The Peutz-Jeghers polyp varies in size from 1 cm to 3.5 cm in diameter, and may be pedunculated or sessile. Since it appears to be composed of non-neoplastic tissue normally found at the site, the Peutz-Jeghers polyp is generally considered a hamartomatous polyp but with an abnormal growth pattern. The most characteristic feature of a Peutz-Jeghers polyp is a central core of smooth muscle that extends into the polyp in an arborizing fashion

(Christmas tree like appearance) and that is covered by either normal or hyperplastic mucosa native to the involved site. Adenomatous & carcinomatous changes have been described in Peutz-Jeghers polyps.[16] The diagnosis of PJS is established by the presence of histopathologically confirmed hamartomatous polyps and at least two of the following clinical criteria: a family history of PJS, the presence of mucocutaneous pigmentation and the presence of small-bowel polyps.[15] Over the years, the standard therapy for Peutz-Jeghers syndrome has been laparotomy and bowel resection to remove symptomatic gastrointestinal polyps that cause persistent or recurrent intussusceptions. However, some patients require multiple surgical resections, which can lead to short gut syndrome. Because of this, it has been recommended that endoscopy be performed to remove all polyps. During each laparotomy, the small bowel should be examined by means of intraoperative enteroscopy (IOE). Nowadays, Double Balloon Enteroscopy (DBE) in combination with capsule enteroscopy are the gold standard for the diagnosis and treatment of the small bowel hamartomatous polyps.[14,15]

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REFERENCES

1. Giardiello FM, Welsh SB, Hamilton SR, Offerhaus GJ, Gittelsohn AM, Booker SV, et al. Increased risk of cancer in the Peutz-Jeghers syndrome. N Engl J Med. 1987 Jun;316(24):1511-4.
2. Calva D, Howe JR. Hamartomatous polyposis syndromes. Surg Clin North Am. 2008 Aug;88(4):779-817, vii.
3. Giardiello FM, Brensinger JD, Tersmette AC, Goodman SN, Petersen GM, Booker SV, et al. Very high risk of cancer in familial Peutz-Jeghers syndrome. Gastroenterology. 2000 Dec;119(6):1447-53.
4. Boardman LA, Thibodeau SN, Schaid DJ, Lindor NM, McDonnell SK, Burgart LJ, et al. Increased risk for cancer in patients with the Peutz-Jeghers syndrome. Ann Intern Med. 1998 Jun;128(11):896-9.
5. Trau H, Schewach-Millet M, Fisher BK, Tsur H. Peutz-Jeghers syndrome and bilateral breast carcinoma. Cancer. 1982 Aug;50(4):788-92.
6. Gruber SB, Entius MM, Petersen GM, Laken SJ, Longo PA, Boyer R, et al. Pathogenesis of adenocarcinoma in Peutz-Jeghers syndrome. Cancer Res. 1998 Dec;58(23):5267-70.
7. Boardman LA, Pittelkow MR, Couch FJ, Schaid DJ, McDonnell SK, Burgart LJ, et al. Association of Peutz-Jeghers-like mucocutaneous pigmentation with breast and gynecologic carcinomas in women. Medicine (Baltimore). 2000 Sep;79(5):293-8.
8. Brosens LA, van Hattem WA, Jansen M, de Leng WW, Giardiello FM, Offerhaus GJ. Gastrointestinal polyposis syndromes. Curr Mol Med. 2007 Feb;7(1):29-46.
9. Davidson NO. Genetic testing in colorectal cancer: who, when, how and why. Keio J Med. 2007 Mar;56(1):14-20.
10. Mehenni H, Blouin JL, Radhakrishna U, Bhardwaj SS, Bhardwaj K, Dixit VB, et al. Peutz-Jeghers syndrome: confirmation of linkage to chromosome 19p13.3 and identification of a potential second locus, on 19q13.4. Am J Hum Genet. 1997;61:1327-34.
11. Peutz JLA. Over een zeer merkwaardige, gecombineerde familiaire pollyposis van de sligmliezen van den tractus intestinalis met die van de neuskeelholte en gepaard met eigenaardige pigmentaties van huid-en slijmvliezen (Very remarkable case of familial polyposis of mucous membrane of intestinal tract and nasopharynx accompanied by peculiar pigmentations of skin and mucous membrane; in Dutch). Nederl Maandschr v Geneesk. 1921;10:134-46.
12. Jeghers H, McKusick VA, Katz KH. Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits; a syndrome of diagnostic significance. N Engl J Med. 1949;241:1031-6.
13. Vesna Živković, Svetlana Pejović, Aleksandar Nagorni, Bratislav Petrović, Aleksandar Petrović, Ivan Ilić. Hereditary hamartomatous gastrointestinal polyposis syndrome. Sci J Faculty Med Niš. 2010;27(2):93-103.
14. Marcela Kopacova, Ilja Tacheci, Stanislav Rejchrt, Jan Bures. Peutz-Jeghers syndrome: diagnostic and therapeutic approach. World J Gastroenterol. 2009 Nov;15(43):5397-408.
15. Michael Manfredi. Hereditary hamartomatous polyposis syndromes: understanding the disease risks as children reach adulthood. Gastroenterol Hepatol. 2010 Mar;6(3):185-96.
16. Karl H. Perzin, Mary F. Bridge. Adenomatous and carcinomatous changes in hamartomatous polyps of the small intestine (Peutz-Jeghers syndrome). Cancer. 1982;49:971-83.

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