Sporadic Case of Peutz-Jeghers Polyp in a 14-Year Boy

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ABSTRACT

Peutz-Jeghers syndrome (PJS) is an autosomal dominant hamartomatous polyposis of the gastrointestinal tract, with pigmentation around lips, the buccal mucosa, and anal area. Patients have a strong family history. Patients of PJS present with abdominal pain, blood in stools, and occasionally melena because of polyps, along with classical mucocutaneous pigmentation. Very rarely a sporadic case of Peutz-Jeghers syndrome occurs in early childhood and adolescent. The case of a 14-year boy is reported, who presented with intussusception and bleeding per rectum due to jejunal polyp and a rectal polyp. Intussusception was treated by resection anastomosis due to vascular impairment. Rectal polyp was removed during colonoscopy. There was no family history of Peutz-Jeghers syndrome or polyps but patient had classical mucocutaneous pigmentation of buccal mucosa. Therefore, this case is of sporadic Peutz-Jeghers polyp (PJP), which is a rare disorder.

Key Words: Melena. Jejunal intussusception. Sporadic Peutz-Jeghers polyp.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disease characterised by hamartomatous polyps in the gastrointestinal tract and mucocutaneous melanin pigmentation and strong family history. The average age at diagnosis is 23 - 26 years. The diagnosis of PJS can be made by using the WHO criteria. The typical histological features are the characteristic frond-like structures, with branching framework of connective tissues and muscles. Sporadic cases of Peutz-Jeghers syndrome, appearing during early childhood and adolescent, can be a difficult problem in diagnosis. Mucocutaneous pigmentation represents the most reliable diagnostic sign. One of the diagnostic features by WHO criteria is any number of polyps and mucocutaneous pigmentation. The authors report a 14-year boy with Peutz-Jeghers polyps causing intussusception, without family history. These cases are important to diagnoses because of the high cancer risk.

CASE REPORT

A 14-year boy was referred from a rural hospital presented in Accidents and Emergency Department, Civil Hospital, Karachi, with complaint of severe abdominal pain, vomiting and bloody stools for the last one month. He had colicky abdominal pain and a significant loss of weight for the past one year. His symptoms dates back to last 5 years but they became severe for the last one year for which he was admitted in one of the hospitals and treated as a case of tuberculosis. He had no family history of similar gastrointestinal disorder or mucocutaneous pigmentation.

On examination, the patient was extremely emaciated, and anemic. He was afebrile; pulse rate was 120/minute with blood pressure of 80/40 mm/Hg and respiratory rate of 30/minute. He had blackish pigmentation on lips, tongue and oral mucosa (Figure 1). Abdomen was tense and tender. On digital rectal examination (DRE), a luminal mass was felt with blood stained current jelly type on finger stall. After admission, he was resuscitated and blood transfusion was given as his Hb was 6 g/dl. His ultrasound abdomen showed thick walled dilated fluid filled bowel loops. X-ray abdomen erect showed multiple air fluid levels in small bowel. CT scan abdomen with contrast showed large intussusception of proximal jejunal loops on right side with intact vascularity and severe edema of wall of intussusceptent. On exploratory laparotomy, he was found to have intussusception of the jejunum and dilatation of duodenum and proximal jejunum. Resection of the intussuscepted segment and jejunojejunal anastomosis was performed as vascularity was compromised. No other gastrointestinal polyp was present.

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found on palpation of the bowel. Postoperative recovery was uneventful. On histopathology, a polyoidal lesion of 5.5 x 4 x 2 cm was found in jejunum. Microscopically, polyp was lined by columnar epithelium with goblet cells; these glands were separated by bands of muscle fibers arranged in an arborizing pattern (Figure 2). There was no evidence of dysplasia or malignancy. The features were of hamartomatous polyps consistent with Peutz-Jegher's polyp.

After few days, colonoscopy was performed and rectal polyp was removed with snare; rest of the colon and terminal ileum was normal. Histopathology was consistent with hamartomatous polyp. Later on, upper GI endoscopy was also performed, which showed mild gastritis but no evidence of polyp up to the duodenum. He comes for regular follow-up, and is doing well. He has been counselled for yearly or 2-3 yearly follow-up for surveillance with upper and lower GI studies, to rule out presence of any cancer.

DISCUSSION

Peutz-Jeghers syndrome appears equally in males and females and is found in all racial groups. The syndrome appears to be mutation of the STK II gene, which is located at 19p13.3 and is autosomal dominant. In 45% of patients there is no family history, may be due to spontaneous mutation.7 Mucocutaneous pigmentation is caused by melanin aggregation and is the characteristic finding of PJS which is present commonly on lips, perioral region, conjunctiva, and buccal mucosa; and sparsely on the fingers, soles of the feet, palms, anal area, penis.5

The Peutz-Jeghers polyp is a true hamartoma with unique histopathologic characteristics. These include the characteristic of frond-like structure. Histologically, Peutz-Jeghers polyps consist of a branching framework of connective tissue and smooth muscle lined by normal intestinal epithelium, rich in goblet cells and the glands having a convoluted shape and arborizing pattern of growth.1 WHO clinical criteria for definite diagnosis of PJS include the presence of two of the following three signs: family history of PJS, mucocutaneous pigmentation or polyposis, mainly of the small-bowel.2 The condition is associated with a substantial risk for adenocarcinoma, mainly of the gastrointestinal (GI) tract.6 It is now known that the gene causing PJS confers for an increase risk of cancer.7

In sporadic cases, because of the absence of involved family members, the characteristic features of polyp or the mucocutaneous pigmentation can be diagnostic.4 In a study including extensive review of clinical and histologic data of 94 polyps meeting WHO criteria, only three patients were labelled as sporadic documented PJPs. The authors suggested that sporadic cases, if exist, are very rare.4 This case had two polyps with PJP characteristics along with classical mucocutaneous pigmentation in oral mucosa. The negative family history suggests sporadic PJPs.

Peutz-Jeghers polyps grow during the first decade of life, and most patients become symptomatic between the ages of 10-30 years.5 This patient had symptoms since he was 10 years of age but his disease was diagnosed in the fifth year of his illness. The presenting complaints of PJS are intestinal obstruction (43%), abdominal pain (23%), blood in the stool (14%), and anal extrusion of polyp (7%). The remaining 13% of cases are diagnosed because of melanin pigmentation.1 The most frequent complication in young age is intussusception occurring in 43% of patients, and is found in 95% of small intestine.5 This patient also presented with jejunal intussusception. PJS patients have a 15.2 relative risk for all cancers and a 93% lifetime risk of any cancer.1 Most authorities recommend surveillance by colonoscopy and upper GI endoscopy.1 Regular small-bowel surveillance in PJS patients is recommended to reduce polyp-related complications, particularly of intussusceptions, and because of the association between PJS and cancer, although there is no data that supports the reduction in risk via surveillance.6 Among the various procedures used for the surveillance of the small-bowel in PJS patients, those that have proven their utility are video capsule endoscopy (VCE), double balloon pushed enteroscopy (DBE), multidetector computed tomography (MDCT), enterography or enteroclysis, and magnetic resonance (MR) enterography.

A negative family history should not deter pursuance of the Peutz-Jeghers diagnosis, since large number of patients described have no family history of mucocutaneous pigmentation or polyposis.3

REFERENCES