CASE REPORTS

Peutz-Jeghers Syndrome

DO Whittle, MG Lee, B Hanchard

ABSTRACT

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant colonic polyposis syndrome. It is a rare condition but is of importance because of the risk of gastrointestinal as well as extraintestinal malignancies and the high penetrance in the family linkage. There has been no report of this condition in the Caribbean. We report a 32-year old male who presents with a history of rectal bleeding on a background history of having colonic polyps. Colonoscopy revealed multiple large colonic polyps with partial obstruction in the descending colon. The histology revealed hamartomatous Peutz-Jeghers polyps. Peutz-Jeghers Syndrome is an important consideration in a young patient with colonic polyps.

Keywords: Hamartomatous, hyperpigmentation, Peutz-Jeghers, polyposis

El Síndrome de Peutz-Jeghers

DO Whittle, MG Lee, B Hanchard

RESUMEN

El síndrome de Peutz-Jeghers (SPJ) se caracteriza por una poliposis colónica autosómica dominante. Se trata de una condición rara, pero de gran importancia debido al riesgo del tracto gastrointestinal, así como las malignidades extraintestinales y la elevada penetrancia genética en los vínculos de familia. No ha habido ningún reporte sobre esta condición en el Caribe. Reportamos aquí el caso de un hombre de 32 años que presenta una historia de sangramiento rectal con antecedentes de pólipos colónicos. La colonoscopía reveló la presencia de grandes pólipos colónicos múltiples, con obstrucción parcial en el colon descendente. La histología reveló la presencia de pólipos hartmartomatosos Peutz-Jeghers. El síndrome de Peutz-Jeghers es una consideración importante en un paciente joven con pólipos colónicos.

Keywords: Hartmartomatosos, hiperpigmentación, Peutz-Jeghers, poliposis

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INTRODUCTION

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant colonic polyposis syndrome associated with mucocutaneous hyperpigmentation (1). It is a rare condition but is of importance because of the risk of gastrointestinal as well as extraintestinal malignancies, and the high penetrance for the intestinal polyps and mucocutaneous hyperpigmentation in the family linkage (2). The polyps are often benign but over time may undergo malignant transformation and surveillance for colonic neoplasia is recommended (3). There has been no report of this condition in the Caribbean.

We present the case of a 32-year old male with multiple large colonic polyps with partial obstruction in the descending colon; the histology revealed hamartomatous Peutz-

From: Department of Medicine and Pathology, The University of the West Indies, Kingston 7, Jamaica, West Indies.

Correspondence: Dr DO Whittle, Department of Medicine, The University of the West Indies, Kingston 7, Jamaica, West Indies. E-mail: dwight.whittle02@uwimona.edu.jm

Jeghers polyps. Peutz-Jeghers Syndrome is an important considera-tion in young patients with colonic polyps.

CASE REPORT

A 32-year old male presented to the Gastrointestinal Clinic, University Hospital of the West Indies, Jamaica, with a 12-year history of intermittent rectal bleeding. He was found to have rectal polyps on proctosigmoidoscopy by a general surgeon about 12 years ago. He had presented at that time with a history of passage of bright red blood per rectum. Barium enema had revealed multiple polyps on the left side of the colon. He defaulted from follow-up for about ten years because he was fearful of having surgery which was suggested to him.

He now re-presents because he has been losing weight and is growing concerned about the polyps. He reports episodes of intermittent passage of bright blood per rectum, not precipitated by constipation or straining. There was no associated pain or tenesmus. Of note, he did not report a significant history of constipation, diarrhoea, nausea, vomiting, jaundice or syncope. He has no family history of polyps and works as an electrical technician.

Examination revealed a young male with evidence of wasting; he was not anaemic, there were no skin changes and there was no lymphadenopathy. His abdomen was flat, non-tender with no masses and on rectal examination, multiple firm rectal polypoid masses were palpated in the rectum. The rest of his systemic examination was normal. Laboratory investigations were all normal, with haemoglobin 14 mg/dL. Colonoscopy revealed multiple polyps in the rectum and sigmoid colon (Fig. 1). At the mid-descending colon, a large polyp prevented further intubation *via* colonoscopy. The polyps were pedunculated and ranged in size from 0.5 to 2 cm. Polpectomies were completed up to the mid-descending colon with removal of approximately 12 polyps. Upper endoscopy revealed a large duodenal polyp (Fig. 2). Barium meal with small bowel follow through revealed no other polyps in the small intestine. Barium enema revealed multiple polyps throughout the colon extending to the ascending colon with partial obstruction at the level of descending colon.

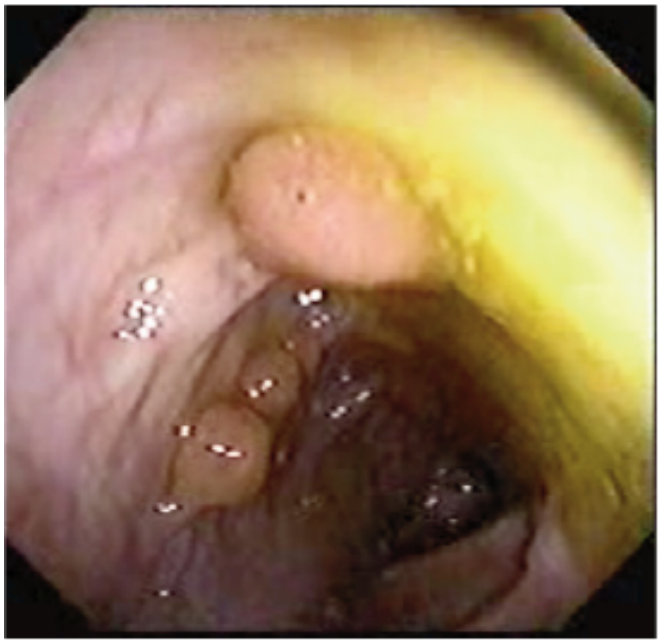

Fig. 1: Polyps in the sigmoid colon.

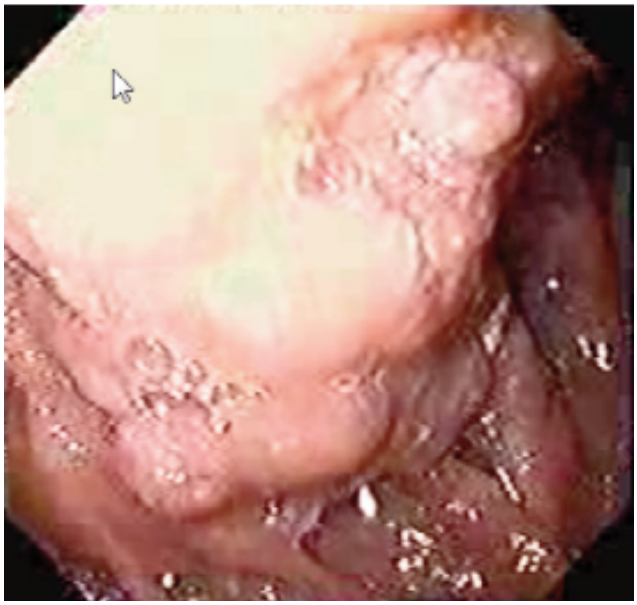

Fig. 2: Polyps in the duodenum.

The polyps histologically were hamartomous polyps typical of Peutz-Jeghers Syndrome. Groups of proliferating glands lined by normal colonic epithelium with abundant goblet cells were the characteristic feature of the polyps, some presenting a sequestered or arborizing pattern due to proliferation of smooth muscle bundles (Figs. 3, 4). No superimposed dysplastic changes were evident.

The patient was advised to have surgical intervention on the basis of the number of polyps in the colon and the risk of complete obstruction at the level of the descending colon.

DISCUSSION

Peutz-Jegers Syndrome has been described in all ethnic groups. It affects males and females equally. There has been no data on the incidence of this syndrome worldwide (4). Peutz-Jeghers Syndrome results from mutations on the STK11/LKB1 (serine/threonine kinase 11) tumour suppressor gene on chromosome 19 p 13.3. (1, 5, 6, 7).

Patients with PJS commonly present with abdominal pain, rectal bleeding, prolapse of tissue through the anus and gastrointestinal intussception with bowel obstruction. The polyps of PJS may ulcerate and bleed and manifest as iron

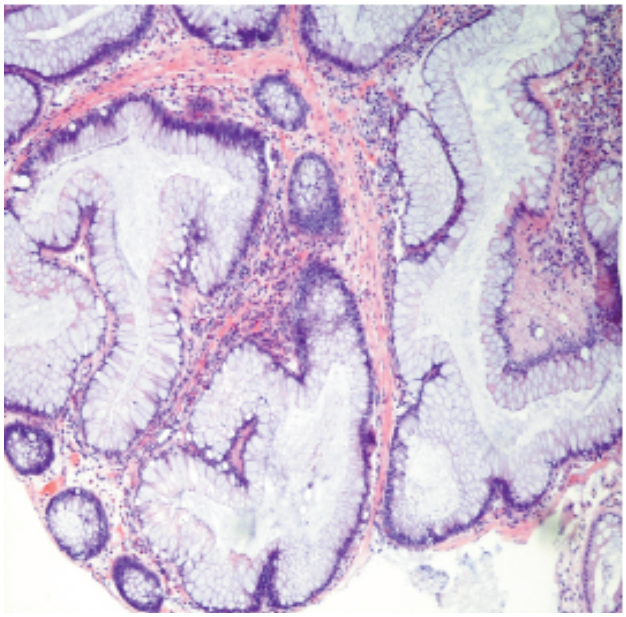

Fig. 3: Histology of colonic polyp (low power).

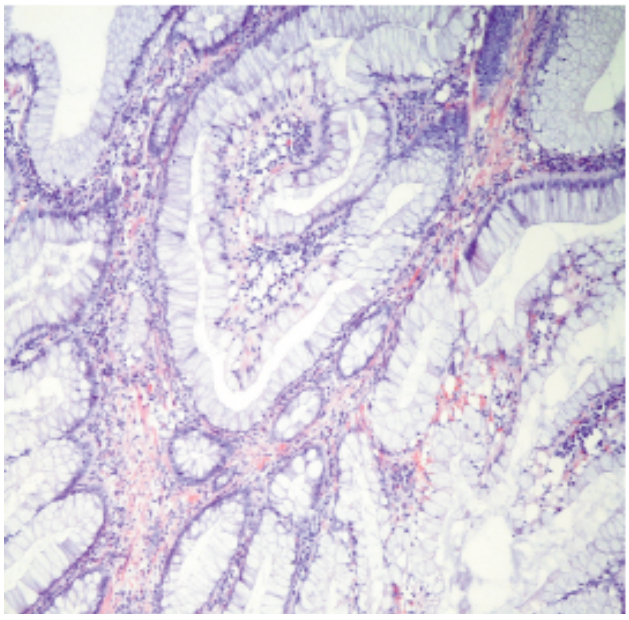

Fig. 4: Histology of colonic polyp (High power).

deficiency anaemia. The index patient had intermittent rectal bleeding for many years but he did not have anaemia.

Mucocutaneous pigmentation and melanin spots are typical of patients with PJS syndrome. They are present in more than 95% of cases and are caused by pigment laden macrophages in the dermis. They appear as small, flat, brown or dark blue spots with an appearance of freckles, most commonly in the peribuccal area but may occur in the intestinal mucosa. They may also be present on fingers and toes, on the volar and dorsal aspect of the hands and feet as well as around the anus and genitalia. These spots may fade after puberty.

Peutz-Jegers Syndrome is a clinical diagnosis that may be supported by genetic testing. For individuals with a histopathologically confirmed hamartoma, a definite diagnosis of PJS requires two of the following three clinical findings: a family history consistent with autosomal dominant inheritance, mucocutaneous hyperpigmentation, or small-bowel polyposis. For individuals without histopathologic verification of hamartomatous polyps, a probable diagnosis of PJS can be made based on the presence of two of the three clinical criteria. For individuals without a family history of PJS, the diagnosis depends upon the presence of two or more histologically verified PJS-type hamartomatous polyps. For individuals with a first-degree relative with PJS, the presence of mucocutaneous hyperpigmentation is sufficient for the diagnosis. The index patient had several hamartomatous polyps involving the colon and duodenum.

Patients who have symptoms or a family history should be screened. Patients should have colonoscopy and polypectomy, upper endoscopy as well as investigations of the small bowel (8, 9). The latter investigations may include capsule enteroscopy, push enteroscopy, intraoperative enteroscopy and double balloon enteroscopy. Imaging of the liver, gall bladder and pancreas should be done as there is an association of PJS with extraintestinal polyps or cancers of these areas.

Regardless of where the polyps occur, the characteristic histology is that of hamartomatous proliferation of complex glands, lined by normal or sometimes hyperplastic epithelial cells. An arborizing configuration is often produced by the bands of smooth muscle in the stroma. In the colon, there may be some variation in the appearance whereby mucosal oedema and inflammation with cystic dilatation of glands create a picture resembling juvenile polyps. The risk of malignancy increases in the presence of dysplastic glands. These are usually seen as elongated hyperchromatic and layered epithelial cells lined by glands and may be low-grade or high grade, the latter equivalent to *in-situ* carcinoma.

All polyps should be removed and sent for histology. Patients should be screened for intestinal and extraintestinal cancers. Annual physical examination, complete blood count, abdominal ultrasound and two yearly endoscopy and colonoscopy are recommended (10, 11). Patients may require surgical intervention with laparatomies and laparoscopies for cancer or complications related to the polyps: small and large bowel intussception, obstruction or persistent bleeding. Forty-eight per cent of patients with PJS develop and die from cancer by age 57 years (9, 12). Patients treated early may have a normal life span.

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