Case Report

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Peutz–Jeghers syndrome: An unusual cause of iron-deficiency anemia

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Abstract:

Peutz–Jeghers syndrome (PJS) is a rare disorder, which is inherited as autosomal dominant. It is characterized by mucocutaneous melanotic spots and multiple hamartomatous polyps with an increased risk of cancer predispositions. PJS can present with complications such as iron-deficiency anemia as a result of bleeding from the gastrointestinal polyps and intestinal obstruction and/or intussusception if the polyps are of large size. We present a case of PJS with iron-deficiency anemia in a 16-year-old boy as a result of occult gastrointestinal bleeding from the hamartomatous gastrointestinal polyps.

Keywords:

Anemia, bleeding, polyp

Introduction

Peutz–Jeghers syndrome (PJS) is a rare disorder inherited as autosomal dominant and characterized by mucocutaneous pigmentations and hamartomatous gastrointestinal (GI) polyps.[1] These patients have an increased risk of development of cancer.[1] About 1 in 50,000–1 in 200,000 live births is reported to have this condition.[2] Genetically it has a germline mutation in SKT11(LKB1) gene which is located at chromosome 19p13.3 encoded for a protein serine/threonine kinase.[3]

The skin in the form of mucocutaneous pigmentations; intestinal systems such as stomach and small and large intestine; and extraintestinal locations such as respiratory tract, gallbladder and biliary tree, pancreas, and urogenital and neurologic systems may all be involved in this syndrome.[1,2] In particular, intestinal polyps can cause iron-deficiency anemia due to bleeding from the GI polyps and can result in intestinal obstruction and intussusception because of their large size.[1] We report a case of PJS in a 16-year-old boy, who presented with severe iron-deficiency anemia.

Case Report

A 16-year-old boy came to the outpatient department with complaints of generalized weakness and loss of appetite for the last 2 years. After clinical and laboratory workup, he was diagnosed to have iron-deficiency anemia and had been on oral iron supplements for the last 1 year. However, there was no improvement of his symptoms. There is no history of any other chronic illness. On general examination, the patient had severe pallor along with melanotic spots in the perioral area, buccal mucosa, and soles of feet since his birth [Figure 1].

On laboratory investigations, his hemoglobin level was 7.5 g/dL. Peripheral blood examination showed microcytic hypochromic anemia [Figure 2]. The fecal occult blood test was positive. His serological markers were within normal limits.

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Endoscopic and colonoscopic examination revealed multiple polyps along the gastrointestinal tract [Figure 3]. An endoscopic biopsy of the polyp from stomach and colonoscopic polypectomy from the ileocaecal junction, transverse colon, descending colon and recto-sigmoid junction was performed.

Histopathological examination of the polyps showed epithelial overgrowth with an arborizing muscle core, which were consistent with multiple hamartomatous polyps [Figure 4]. Thus, a diagnosis of PJS was rendered.

At 6-month follow-up, the patient is doing well with a significant improvement in his symptoms.

Discussion

Initially described by Peutz and Jeghers in the years 1921 and 1949, respectively, PJS is a rare hereditary autosomal dominant condition.[1] Clinically, most striking feature is the presence of mucocutaneous melanotic spots and GI polyps.[1-3]

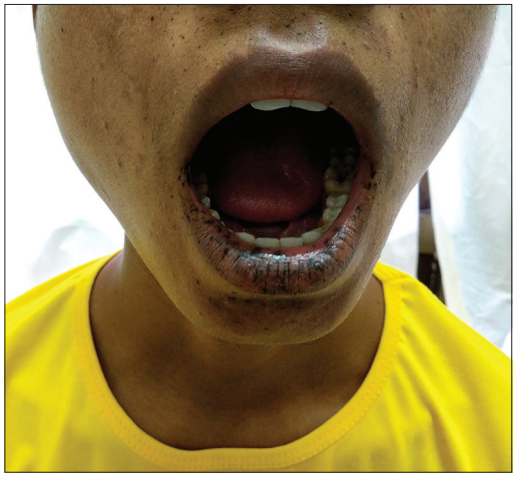

Figure 1: Melanotic spots in the perioral area

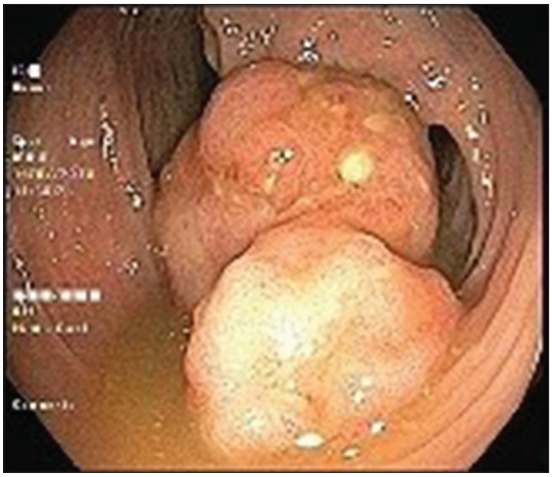

Figure 3: Multiple polyps in transverse colon

The skin lesions are commonly seen around the mouth, nostrils, fingers and toes, and perianal and genital areas.[1] Mostly found in infancy, these skin lesions can be seen in 93% of patients.

The polyps are usually located along the gastrointestinal tract most commonly at small intestine (60%–90%) and large intestine (50%–64%).[4] Rarely, these polyps can also be found in the extraintestinal locations. Histologically, PJ polyps are hamartomatous polyp which is characterized by extensive arborizing smooth muscle proliferation along with frond-like epithelial hyperplasia and cystically dilated glands invaginating into the submucosa or muscularis propria.[1]

PJS patients may have complications such as iron-deficiency anemia as a result of occult bleeding from the intestinal polyps.[1,3] The present case also had iron-deficiency anemia as a result of occult bleeding from the GI polyps. Therefore, a thorough systemic

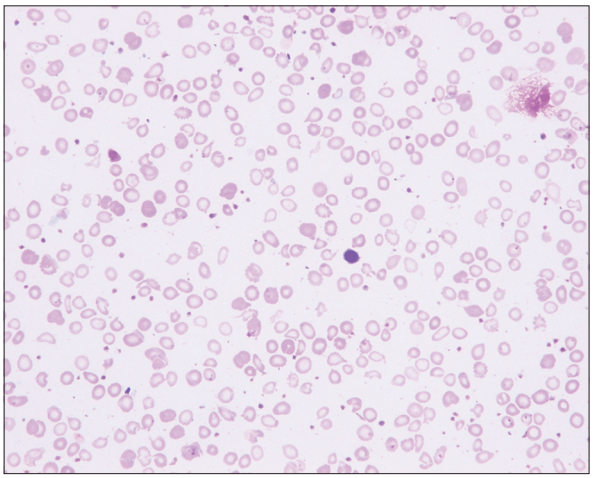

Figure 2: Peripheral smear showing microcytic hypochromic red blood cells (Leishman, ×40)

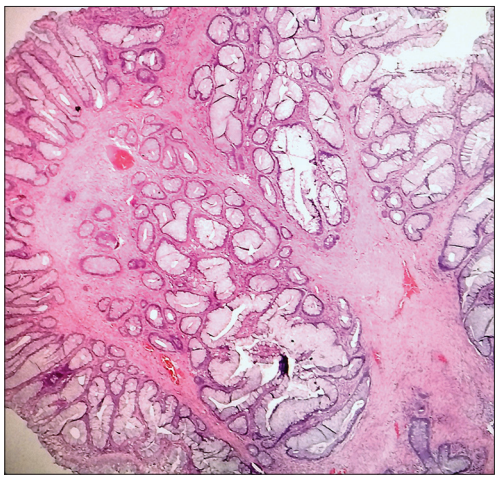

Figure 4: Histopathology of polyp showing epithelial overgrowth with an arborizing muscle core (H and E, ×40)

examination and a detailed laboratory workup in cases of iron-deficiency anemia should be done, especially in children.[3] This is of particular importance in developing countries, where the prevalence of nutritional anemia such as iron-deficiency anemia is high.[5]

If the PJ polyps are large, they can cause obstruction and/or intussusception.[1] However, the greatest risk is the development of malignancy in about 50% of these patients.[1] The median age of development of malignancy is about 50 years.[1] Because of the malignant potential, PJ polyp should be removed endoscopically or surgically.[6] In the present case, the polyps were endoscopically resected.

PJS patient needs early diagnosis as anemia and intestinal complications can increase the morbidity. Moreover, these patients have high risk of developing malignancy. In developing countries, the prevalence of nutritional anemia such as iron-deficiency anemia is high; however, the characteristic association of mucocutaneous pigmentation along with gastrointestinal symptoms clinches the diagnosis of PJS.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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