The importance of early diagnosis and surveillance in Peutz-Jeghers Syndrome: A case report

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Introduction: Peutz-Jeghers syndrome is a rare autosomal dominant inherited disorder characterized by hamartomatous intestinal polyps and mucocutaneous pigmentation. Most cases appear to be linked to the mutation of the STK11 gene. Patients are at a lifetime risk of gastrointestinal and non-gastrointestinal cancers. Case Presentation: The present study offers the case of this rare disorder in a young woman revealed by jejunal obstruction caused by intussusception. A 32-year-old woman was referred to the surgical department with symptoms suggestive of an obstructive syndrome. On examination, there were multiple perioral pigmented lesions. An urgent exploratory laparotomy revealed bowel obstruction caused by an intussusception with a large polyp. The patient suffered another similar episode 4 years before leading to the diagnosis of Peutz-Jeghers Syndrome, however she was under no surveillance. Patients with pigmented lesions and a family member suffering from the mentioned syndrome should perform endoscopy and genetic tests to diagnose early and avoid complications.

Conclusion: Peutz-Jeghers Syndrome is difficult to treat due to its nonspecific symptomatology and late diagnosis. Life-threatening complications such as intussusception and various types of cancer are unanticipated. It is vital to diagnose and perform routine screening, which will make it possible to prolong the survival of many patients.

Keywords: hamartomatous polyps, anemia, intussusception, screening, family history

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mangioma, and Computer Tomography (CT) showed a benign pulmonary tumor located in the left inferior lobe.

Later in October 2010, the patient was presented to the gastroenterology department for obscure gastrointestinal bleeding. CT enteroclysis had shown several jejunal polyps. A spiral endoscopy procedure was performed. Multiple pedunculated and sessile polyps of varying sizes in the small bowel were detected beyond the ligament of Treitz, some with superficial erosions, and friable (Figure 1). A hot snare polypectomy was performed for several lesions (Figure 2). Four polyps were removed from the proximal jejunum (Figure 3), two of which were PJS polyps with high-grade dysplasia and in situ carcinoma, emphasizing the risk of gastrointestinal cancer.

The Regional Gastroenterology and Hepatology Institute has managed the patient from 2010. In present, she is treated endoscopically every two years with polypectomy. Polyps larger than 15-20mm must be resected within elective polypectomy to prevent obstruction and intussusception. Smaller polyps should be resected as well if they are symptomatic. Consecutive to a routine endoscopy surveillance and polypectomies for the last 13 years the physicians were able to prevent complications.

Another essential and frequent complication is the bleeding from polyps which can be manifested by anemia, melena, or rectorrhagia. Occult hemorrhages can be revealed by a complete blood count and a hemoccult test every year. Also, the anemia is managed with iron supplements.

As for routine screening for non-GI cancer, the most important for a female patient is the breast and gynecologic exam every year. The breast exam consists of MRI, mammography, and ultrasonography. Moreover, the gynecologic exam should include PAP and HPV tests and screening for ovarian and uterine cancer [2].

**Discussions**

Most cases of PJS are revealed at a young age, either by acute intestinal obstruction or anemia. The present study offers the case of this rare disorder in a young woman revealed by jejunal obstruction caused by intussusception. The clinical picture suggests the diagnosis of Peutz-Jeghers syndrome. Mucocutaneous pigmented lesions represent the first clue of the diagnosis, as they occur in approximately 95% of patients with PJS [5]. Patients with perioral or buccal pigmentation and/or ≥ 2 GI hamartomatous polyps or a family history of Peutz-Jeghers syndrome should be evaluated for this syndrome, including testing for STK11 mutations [6].

PJS might be sporadic or inherited, and clinical manifestations are impressively heterogeneous in both forms. The family history of PJS was detected in the present case suggesting a link with the STK11 gene. Approximately 60-78% of individuals with PJS have an affected relative [7]. The risk of passing the abnormal gene from an affected parent to an offspring is 50% for each pregnancy. The risk is the same for males and females [7]. Other genetic abnor-
malities have been identified, including over-expression of COX-2, mutations in 19q13 gene, in 6p11 gene, and in the MYH11 gene [6].

The STK11 gene produces a protein that is involved in the regulation of cell division and programmed cell death (apoptosis). It also interacts with p53, a major tumor suppression protein. Pathogenic mutations in STK11 lead to either cessation or dysfunction of protein production by the gene and uncontrolled cell growth, which can lead to the development of benign polyps (hamartomas) and cancer [7].

Predisposition to cancer in multiple organs constitutes an essential feature of this condition [6]. Ishida H et al. have demonstrated that the most frequent site of the malignant tumor after the gastrointestinal tract (colo-rectum, small intestine including the duodenum, and stomach), was the breast, followed by gynecological organs (uterus and ovary), pancreas, and lung [8].

The dark pigmented spots (melanocytic macules) are thought to be caused by inflammation and blockage of melanin migration from cells where it is produced (melanocytes) to cells forming the outermost layer of the skin (keratinocytes) [7]. The pigmentation is frequent in patients and might be the first clinical sign encountered by a physician [4]. It is imperative to diagnose PSJ as soon as possible especially at a young age and in patients with de novo mutations with no family history as to avoid multiple laparotomies, bowel resections and small bowel syndrome subsequently.

European Society of Gastrointestinal Endoscopy recommends a baseline esophagogastroduodenoscopy and colonoscopy at the age of 8 years in asymptomatic individuals with Peutz–Jeghers syndrome [2]. The interval for screening endoscopy is between 1–3 years based on phenotype for esophagogastroduodenoscopy and colonoscopy [2].

This case emphasizes the importance of patient education, routine surveillance, and tertiary prevention strategies to avoid complications in individuals diagnosed with PJS as well as the identification and screening of other family members that might have the same condition.

Conclusion
PJS is a pathology challenging to treat due to the nonspecific symptomatology and late diagnosis. Rare, life-threatening complications of PJS, such as intussusception, bleeding, and various types of cancer, are unanticipated. Thus, it is of utmost importance to diagnose PJS as soon as possible and perform routine screening, which will make it possible to prolong the survival of many patients. The key aspect of clinical diagnosis in PJS is the perioral mucocutaneous pigmentation encountered in most patients.

Author contribution
MGB (Conceptualization, Data Curation, Formal Analysis, Investigation, Writing-original draft, Writing-review and edit)

Conflict of interest
None to declare.

References