Unexpected Peutz-Jeghers Syndrome in an Adult Presenting with Intermittent Upper Intestinal Obstruction. A Case Report

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INTRODUCTION

Peutz-Jeghers syndrome (PJS) is an autosomal dominant inherited disease, belonging to the hamartomatous polyposis syndromes [1]. It is characterized by multiple hamartomatous polyps of the gastrointestinal tract associated with oral and anal mucocutaneous pigmentations [2]. The genetic lesion responsible for the phenotypic expression is in 66-94% of the cases considered to be a mutation on the short arm of chromosome 19 (19p13.3) of STK11/LKB1. Abrogation of this tumor suppressor gene results in both polyposis along with probably a separate process, leading to tumorigenesis, and thus an increased risk of developing malignancies [3, 4]. Penetrance of the gene mutation is variable, resulting in a spectrum of phenotypic manifestations (inconsistent number, localization of polyps, different presentation of the macules) [5].

CASE REPORT

A 41-year old woman without any past medical history, besides 2 deliveries and 3 abortions, was admitted to our hospital complaining of intermittent episodes of nausea, vomiting and pain in the right upper abdominal quadrant and epigastrium. The symptoms had no relation with food intake. Bowel transit was normal with stools every 1-2 days. The presenting symptoms started about a year prior to presentation and were partially ameliorated by antacids, antispasmodics and pain relievers. A weight loss of 8 kg in this year was noted but with preserved appetite and periods of regaining weight. The patient came from a rural area, worked in a botanical garden and denied any exposure to environmental or professional toxics.

The clinical exam revealed a slim woman with a BMI of 20.7kg/m², pale, with dry mucosae, café-au-lait plaques on the skin of the upper trunk (similar to pityriasis versicolor). The abdomen was elastic, mobile with respiration, presenting tenderness in the epigastrium and the right upper quadrant.

Laboratory data showed a mild iron deficiency anemia (hemoglobin 11.2g/dl), hypertriglyceridemia, hypocalcemia and a moderate inflammatory syndrome. No other blood, urine or stool changes were detected.

Esogastroduodenoscopy revealed a gastric atrophic mucosa with micropolyps of 0.2cm (Fig.1), multiple erosions.
in the second part of the duodenum (DII) and an important duodeno-gastric reflux. *Helicobacter pylori* was negative. The same aspect had been found several months before in another service.

The abdominal ultrasound discovered a mass in the right upper quadrant that was suggestive for an entero-enteral intussusception with a circumferentially thickened wall (10mm), raising the suspicion of a giant polypoid tumor in the small bowel (Fig. 2). Bowel changes were also found in the right and left iliac fossa. The Doppler examination identified a possible tumoral mass of 20/20mm, vascularized through a branched pedicle, and considered to be part of the proximal jejunum. The bowels had normal peristalsis and parietal vascularization. No sign of ischemia was identified. These findings were confirmed by contrast enhanced ultrasound examination.

The abdominal computer tomography (CT) described the intestinal intussusception of the first jejunal loop (size 149/60/78mm) but did not bring any additional information to the underlying cause (Fig. 3). Small mesenteric adenopathies were present.

A spiral enteroscopy was considered. Inspection of the entire jejunum identified two polyps. The first lesion was located at the junction of DIII with the first jejunal loop, it had about 40mm and was the cause of the invagination (Fig. 4).

Another 40mm polypoid tumor with ulcerations was found in the terminal jejunum. The procedure was both diagnostic (multiple biopsies taken from the polyps) and therapeutic, performing reduction of the intussusception. Additionally, colonoscopy was performed and no lesion was found. Tumor markers were checked: CEA was normal, but CA125 was elevated (101.6 IU/ml).

Immediately after enteroscopy the patient had no complaints, but the day after the procedure, symptoms of small bowel obstruction (colicative pain in the right upper quadrant, nausea and vomiting) appeared. The patient was transferred to the surgical department. A double small bowel segmentary resection with termino-terminal anastomosis was performed (Fig. 5). Considering the findings, an intestinal polyposis syndrome was suspected and was sustained by the morphological description of the lesions as being hamartomatous polyps (Fig. 6). We add here that the first two histological exams, the gastric biopsy and the jejunum biopsy described nonspecific chronic inflammation.

In search of mucocutanous lesions, an isolated oral pigmentation was found on the mucosa of the lower lip. No other lesions could be identified in the oral and anal region or on the hands and feet.

A further detailed family history was obtained. The patient had no recollection of her parents or brother having any gastro-
Peutz-Jeghers syndrome, also known as hereditary intestinal polyposis syndrome is diagnosed according to the WHO criteria when the presence of mucocutaneous pigmentation, intestinal pathology, but her older sister had been investigated for colonic polyps.

The patient had an uneventful recovery and remained well 3 months later. Her asymptomatic children were also evaluated. They presented clinical features of PJS (dark macules around the oral area) and similar gastric micropolyposis. No polyps were found in the first part of the small bowel that was evaluated endoscopically. The patient and the children will be clinically and biologically reevaluated on a specific schedule. In the meantime a gynecological surveillance has been suggested and is mandatory, taking into consideration the elevated CA125 and the association between PJS and gynecological cancers.

**DISCUSSION**

Peutz-Jeghers syndrome is highly penetrant with variable expression. Only one case of non-penetration of an LKB1 mutation has been reported so far [18]. De novo mutations are described as well [5]. Moreover, variability in the timing and number of mucocutaneous pigmentations, polyps and cancer is highly unpredictable [6]. This is why, even if genetic testing is usually not necessary [6], analysis of germline STK11/LKB1 mutations may be necessary in any case of PJP which does not fulfill the WHO criteria.

**CONCLUSION**

Patients with PJS should be followed by a multidisciplinary team. Patients should be educated on the potential symptoms of intestinal obstruction and instructed on the need for cancer surveillance. Counseling and testing of asymptomatic family
members is mandatory in order to prevent complications, including malignancy.

Conflicts of interest: None to declare.

REFERENCES