

Peutz-Jeghers syndrome associated with intestinal obstruction due to intususception, a rare disease with a frequent complication. Case report

Síndrome de Peutz-Jeghers asociado a obstrucción intestinal por intususcepción: una rara enfermedad con una complicación frecuente. Presentación de caso

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Summary

Peutz-Jeghers syndrome, is an autosomal dominant neoplastic syndrome characterized by the formation of hamartomatous polyps along the gastrointestinal tract, typical mucocutaneous pigmentations, and an elevated lifetime risk of cancer. We present the case of a 21-year-old woman with a known history of Peutz-Jeghers syndrome, without follow-up, who presents symptoms of abdominal pain, nausea and emesis with intolerance to oral intake, with a tomographic study and upper Gi series that lead to the diagnosis of small bowel obstruction with intussusception at the level of the second portion of the duodenum. This article reviews the literature and the radiological features of Peutz-Jeghers syndrome.

Resumen

El Peutz-Jeghers es un síndrome neoplásico autosómico dominante que se caracteriza por formar pólipos hamartomatosos a lo largo del tracto gastrointestinal, pigmentaciones mucocutáneas típicas y un riesgo elevado de cáncer de por vida. Se presenta el caso de una mujer de 21 años de edad con historia conocida de síndrome Peutz-Jeghers, sin seguimiento, quien presenta clínica de dolor abdominal, náuseas y emesis con intolerancia a la vía oral, con estudio tomográfico y serie gastroduodenal que llevan al diagnóstico de obstrucción intestinal con intususcepción en la segunda porción del duodeno. En el presente artículo se revisan la bibliografía y las características radiológicas del síndrome de Peutz-Jeghers.

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Introduction

Peutz-Jeghers syndrome (PJS) is an autosomal dominant neoplastic disease with hamartomatous polyps along the gastrointestinal tract, characteristic mucocutaneous pigmentations and an elevated lifetime risk of cancer (1).

The first descriptions of this disorder date back to the late 1800s by Dr. Conner and later in the 1920s by Dr. Peutz. However, a combination of intestinal polyposis and mucocutaneous pigmentations was first described as a distinct entity in 1949 by Dr. Jeghers (2). The estimated incidence of PJS ranges from 1:50,000 to 1:200,000 births with no racial or sex predilection (3).

Most cases result from a mutation in the STK11 tumor suppressor gene; as a result, PJS is associated with an increased risk of malignancies resulting in significant morbidity and mortality. The most common cancers in this patient population include gastrointestinal, genitourinary, breast, and lung malignancies (4).

A clinical diagnosis of PJS is made when one of the following criteria is met:

1. Two or more pathologically confirmed Peutz-Jeghers polyps. Histologically, PJS polyps of the small intestine are characterized by a distinctive papillary villous architecture with a tree-like arborization of compact bundles of smooth muscle within the lamina propria and a relatively normal overlying

epithelium. However, colonic polyps of PJS can mimic mucosal prolapse polyps or virtually any prolapsing colonic polyp (5).

2. Any Peutz-Jeghers polyp and family history of PJS.
3. Mucocutaneous pigmentations with a family history of PJS. 90% of patients with PJS present a characteristic hyperpigmentation of the skin and mucous membranes as macules ranging from 2 to 12 mm in diameter, dark brown, bluish-brown or blue depending on the racial type. They are preferentially located on the lips (96 %) with predominance of the lower lip and on the oral mucosa (83 %). The gums and palate may be affected, while the tongue and jaws are generally free of lesions. The eyelids are usually affected (36%), and less frequently the conjunctiva may be involved. On the face, lesions tend to converge around the mouth and nose. Pigmentation in the intestinal mucosa is observed in the same way as the skin in endoscopic studies (6).
4. Any Peutz-Jeghers polyp in patients with characteristic mucocutaneous pigmentations.

In addition, genetic testing, detailed clinical history (including detailed childhood information) and family history play a key role in the diagnosis. Relatives of confirmed cases of PJS should undergo genetic testing (7).

Case report

21-year-old woman with known history of PJS due to histopathology of polyps previously resected by endoscopy and characteristic skin lesions, without follow-up. She presented with abdominal pain, nausea and oral intolerance. Computed axial tomography (CT) with intravenous contrast medium shows invagination of a long segment of the proximal jejunum to the second portion of the duodenum, causing complete obstruction with consequent dilatation of the proximal duodenum and stomach. Coronal CT showed the intussusception mass with displacement and obstruction of the bile duct (Figure 1). Additionally, duodenal, jejunal, ileal and colonic polyposis was observed, compatible with PJS (figure 2).

An esophagogastroduodenal series with water-soluble iodinated contrast medium was performed orally due to the suspicion of possible perforation, and because upper gastrointestinal endoscopy (EGD) was to be performed later. The series shows loss of the usual morphology with marked dilatation of the proximal duodenum and loss of the connivent valves; in the second portion of the duodenum there is a focal decrease in its caliber, and there is no evidence of passage of contrast medium to the third and fourth portion of the duodenum. Likewise, ovoid images with negative filling defect are observed in the gastric body and first portion of the duodenum corresponding to polyps (figure 3).

In the EGD, countless polyps are evidenced in the stomach and duodenum. Samples were removed for histopathological study, confirming the diagnosis of hamartomatous polyps. At the intussusception level, reduction maneuvers were performed to reestablish intestinal flow, and polyp resection was achieved (Figure 4). The patient had an adequate clinical evolution, with no post-procedure complications, and was discharged from the hospital.



Figure 1. CT with intravenous contrast medium. a) axial section and b) coronal reconstruction. Invaginated intestinal mass (red arrow) of the proximal segment of jejunum in the duodenum, which completely obstructs the proximal duodenum and retracts the biliary tract, causing dilatation of the stomach (arrow head) and the extrahepatic biliary tract (asterisk).

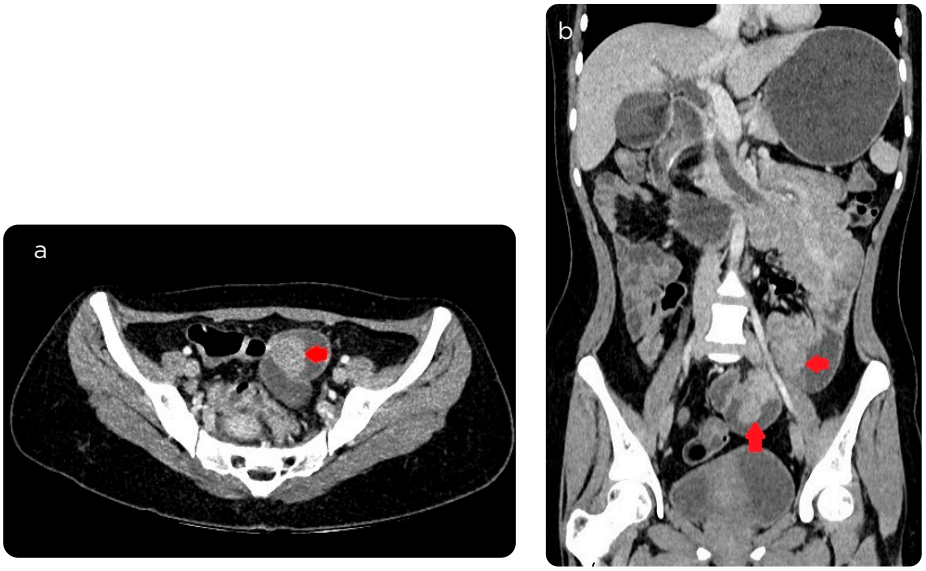


Figure 2. CT with intravenous contrast medium. a) axial section. b) coronal reconstruction. There are multiple prominent polypoid lesions, which enhance with contrast medium, located in jejunum and ileum (red arrows), compatible with PJS.

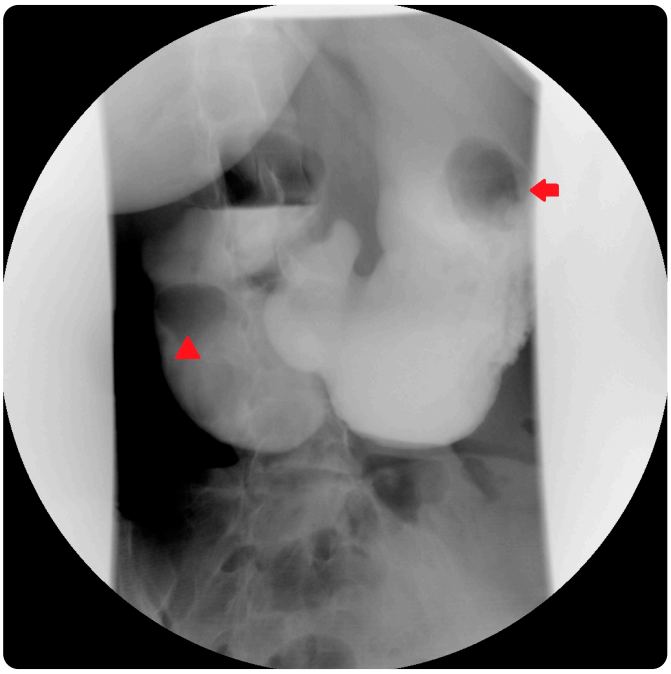


Figure 3. Esophagogastroduodenal series with water-soluble iodinated contrast medium, static image at the level of the upper abdomen. Negative filling defects, prominent ovoid, located in the gastric body (red arrow) and the first portion of the duodenum (arrow head), compatible with polyps.

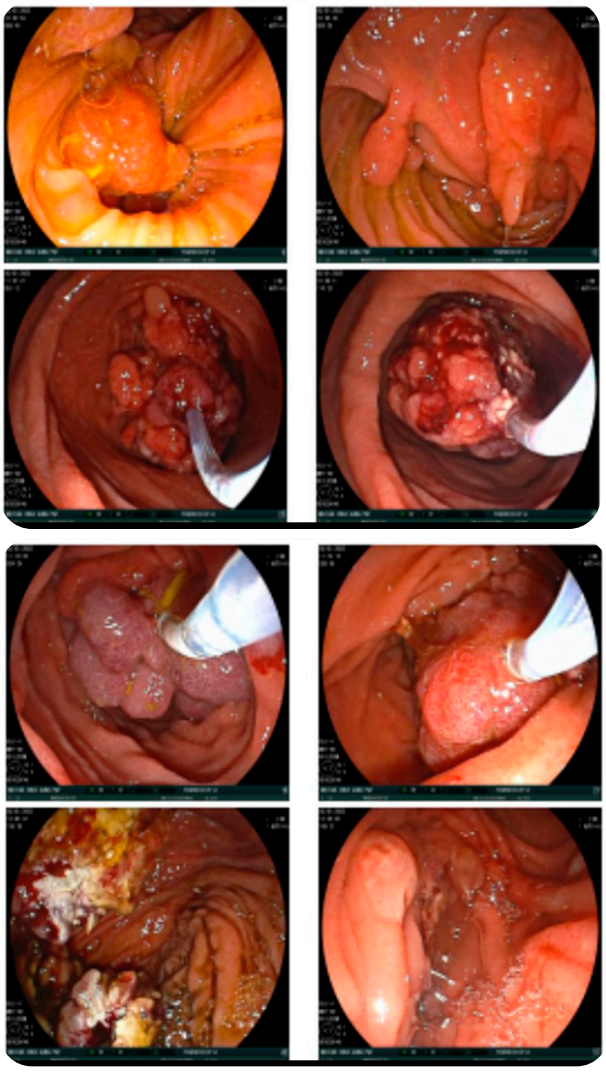


Figure 4. Endoscopic study of the upper gastrointestinal tract showing multiple polyps in the stomach and duodenum. During the study it was possible to resect these lesions, including the one located at the site of intussusception, with subsequent recovery of intestinal flow. The pieces were sent for histopathological study with a report of hamartomatous polyps.

Discussion

CT plays a vital role in the follow-up of these patients, particularly in the evaluation of the small bowel. Polyps are easily visualized as low-density masses, which show mild to moderate and homogeneous enhancement after contrast medium administration, without infiltrative signs. CT provides the necessary information on the exact location and dimension of the masses, the relationship to surrounding tissues and the presence of concomitant malignant lesions.

On the basis of the different features, CT can help distinguish PJS polyps from other intestinal neoplasms, such as small bowel adenocarcinoma, ampulla of Vater tumors, duodenal polyp, lymphoma, carcinoid tumor, stromal tumors and lipoma (3).

The occurrence of multiple polyps in the stomach, small intestine and colon is not limited to PJS; other polyposis syndromes, such as familial adenomatous polyposis, juvenile polyposis, Cowden syndrome and Cronkhite-Canada syndrome, should be excluded. Juvenile polyposis, particularly the generalized form involving the stomach, small intestine and colon, is very likely to mimic PJS, due to the young age of the patients and the pedunculated nature of the larger polyps (4). In most cases, the combination of radiological findings and the clinical presentation of the patient will suggest the actual diagnosis. However, definitive diagnosis always depends on adequate histologic examination of several polyps; when a solitary Peutz-Jeghers polyp is seen, the diagnosis may be more complicated. Such lesions are likely to be mistaken for an isolated juvenile polyp or other polypoid mucosal mass, such as a villous adenoma or polypoid adenocarcinoma in any area. The lobulated surface of the PJS polyp contributes to this confusion (8).

Intussusception is a frequent complication of Peutz-Jeghers polyps, mainly in early life. It occurs in up to 47% of patients, and is mostly (95%) due to polyps located in the small bowel, predominantly ileal or jejunal (9, 10). About 69% of patients experience some episode of intussusception during their lifetime (11). The mean age of intussusception in PJS is 15 years, but with a wide variability (range: 3.7-45.4 years) (9). In pediatric PJS, approximately 30 % of patients required laparotomy before the age of 10 years and 68 % before the age of 18 years. Seventy percent of the initial laparotomies were performed urgently due to intestinal obstruction (12). In PJS intussusception is usually limited to the small bowel and is visualized with antegrade studies. Because intussusception is often not associated with bowel obstruction, imaging plays a critical role, especially CT. Careful observation will identify the intussusception as an area of narrowed bowel (intussusceptum) traveling through a coiled loop of surrounding bowel (intussuscipiens). In most cases, it is also possible to identify the polyp leading to intussusception. Colonic or ileocolic intussusception is not very frequent, and its identification is easier after administering a retrograde barium enema.

The cumulative risk of developing cancer in patients with PJS aged 15-64 years ranges from 37% to 93% (13). Colorectal cancer is the most common gastrointestinal malignancy in patients with PJS, and the risk of developing colorectal cancer is as high as 39%, which increases with age. In this context, in the presence of colonic intussusception, neoproliferative growth should be suspected as a first possibility (14).

Ultrasonography and CT are the modalities of choice for evaluation of suspected intussusception, and will show the multiple layers of the intussusceptum and intussuscipiens. The typical image is the concentric layered morphology known as "bull's-eye" or "doughnut" on transverse slices for ultrasound and axial for CT, and pseudorhinal morphology on longitudinal slices for ultrasound and coronal or sagittal slices for CT. It is key to detect the fat and vessels of the invaginated mesocolon, which simulate the renal hilum, while the edematous intestinal wall resembles the renal parenchyma. Another characteristic is the crescent shape with echogenicity and/or attenuation of fatty tissue that represents the invaginated mesenteric adipose tissue observed in ultrasound or CT images, respectively (15).

PJS is a relatively rare condition and requires a specialized approach to manage the elevated risks of malignant tumors and complications related to gastrointestinal polyps. The radiologist plays a key role in the timely diagnosis of these patients, and should have a high degree of suspicion when reviewing studies of patients with a diagnosis or family history of Peutz-Jegher syndrome. It is now widely recognized that polyp size is the most important risk factor for intussusception with small bowel obstruction and that this is usually due to polyps $\geq$ 15 mm in diameter. Consequently, large (10-15 mm) or symptomatic or rapidly growing polyps should be removed (16).

Imaging surveillance protocols are not yet fully defined, given the low incidence of this disorder. However, continuous surveillance of patients with PJS may prolong life expectancy and improve outcomes through early detection of neoplasia.

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