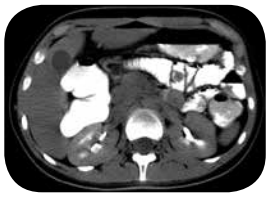

Gastrointestinal Hamartomatous Polyposis with Intestinal Intussusception in a Patient with Peutz-Jeghers Syndrome: A Case Report

Poliposis hamartomatoso gastrointestinal con intususcepción intestinal, en paciente con síndrome de Peutz-Jeghers: Presentación de un caso

Key words (MeSH)

Peutz-Jeghers síndrome
Multidetector computed tomography
Intestinal neoplasm
Colonic polyps

Palabras clave (DeCS)

Sindrome de Peutz-Jeghers
Tomografía computarizada multidetector
Neoplasias intestinales
Pólipos del colon

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Summary

This article presents the case of a 17-year-old female patient with abdominal pain following an endoscopic procedure (polypectomy). The patient has a known diagnosis of Peutz-Jeghers syndrome (PJS), which is manifested by hamartomatous intestinal polyposis and mucocutaneous hyperpigmentation. This review presents the tomographic characterization of multiple gastric and intestinal polyps, including intussusception, which is one of the main complications in these patients.

Resumen

Se trata del caso de una paciente de 17 años de edad, quien consulta por dolor abdominal posterior a un procedimiento endoscópico de resección de pólipo intestinal. La paciente tiene diagnóstico conocido de síndrome de Peutz Jeghers (SPJ), el cual se manifiesta por poliposis intestinal de tipo hamartomatoso e hiperpigmentación mucocutánea. En esta revisión se describen los principales hallazgos del SPJ en tomografía multicorte del abdomen, incluyendo la intususcepción que es una de las complicaciones más frecuentes de estos pacientes.

Clinical Case

A 17-year-old patient, female, with antecedent of Peutz-Jeghers syndrome (PJS) diagnosed at 6 years of age and inherited by paternal line. Father has antecedent of PJS and history of hemicolectomy. The patient has characteristic mucocutaneous hyperpigmentation and known gastrointestinal polyposis. In the usual follow-up of the patient, balloon enteroscopy and colonoscopy was performed. During the anterograde endoscopic procedure, multiple gastric polyps were observed. During the endoscopy there was traumatic fragmentation at the passing of the equipment, which triggered transient bleeding of one of the gastric polyps. Subsequently, polypectomy in the proximal jejunum was done. Afterwards, a retrograde colonoscopy was performed; in the progression to the mid-distal ileum, a giant polyp was identified, which was marked with chinese ink for scheduled surgical removal. Additionally, polypectomy was performed on distal and blind ileum.

During her recovery the patient had vomiting with hematological remains and diffuse abdominal pain. After recovery, the patient was discharged. The next day she required assessment in emergency service for new episodes of vomiting and abdominal pain. Up to physical exam the patient is hemodynamically stable, with abdominal distension, defense and pain to superficial palpation of the abdomen with hemoglobin of 12.2 mg/dl and hematocrit of 36.5 %. Due to suspicion of hollow viscera perforation a multi-slice computed tomography (MCT) of the abdomen was performed. The study was performed on a multi-slice tomograph (Toshiba Asteion) with 4-row detectors, with collimation (slice thickness) of 3 mm; She was given 800 cm^3 of diluted water-soluble iodinated con-

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trast medium oral solution and 60 cm^3 of non-ionic intravenous iodinated contrast medium. Portal (60 seconds) and late phases of the abdomen were obtained (3 and 10 minutes).

In the MCT of the abdomen multiple intraluminal polypoid nodular lesions were identified with soft tissue density - due to known hamartomatous polyps in the patient -, which compromise the stomach, jejunum and ileum (Figure 1). Signs of ileal-ileal intussusception with intestinal polyp at the head of the intussusception located in the mid-distal ileum are observed, without signs of intestinal obstruction (Figure 2).

Because no signs of viscera perforation were found nor of intestinal obstruction and to clinical resolution of the symptomatology, expectant management within the hospital for 48 hours, with blood count control with Hb of 11.1 g/dl and HTO Of 34.2 % was determined. She presented good evolution and tolerance to the oral delivery, for which medical discharge was given.

Two weeks later, surgical resection of the polyp of the ileum; associated intussusception was found in the mid-distal ileum with a segment of at least 20 cm containing the previously marked polyp, without intestinal obstruction. 15 cm were resected including the described polyp, which was sent for pathological study and end-to-end anastomosis was done (figure 3).

The patient was hospitalized for three days with favorable evolution, so she was discharged. Five days later the control by external consultation was done. The patient was asymptomatic.

Topic revision

Peutz-Jeghers disease is a family condition, described for the first time by Hutchinson in 1896, characterized by the presence of polyps in the gastrointestinal tract (predominantly in the small intestine) of hamartomatous type and mucocutaneous hyperpigmentation. It appears similarly in men and women, with an approximate prevalence of 1/100 000 and without ethnic predominance (1). This disease is caused by a mutation in the gene STK 11 on chromosome 19p. The PJS is autosomal dominant with almost complete penetrance.

In these patients the risk of intestinal and non-intestinal cancer increases, with a cumulative risk for any neoplasia between the ages of 15 and 64, greater than 90 %. The most common neoplasm is located in the small intestine and has a relative risk (RR) of 520, followed by stomach (RR 213), colon (RR 84) and esophagus (RR 57). The most frequent extraintestinal tumors are breast and pancreas (2).

Clinically, it manifests with oral, anal and skin mucosal hyperpigmentation, (hands, feet, genitals) from birth. Hyperpigmentation plus gastrointestinal polyps determine the diagnosis of the syndrome (3). The histology of polyps may also guide diagnosis, as they are characterized by a central region of smooth muscle that originates in the muscularis mucosae and branches through the polyp which is covered by a hyperplastic epithelial layer (4). Commonly, patients consult for complications secondary to thepresence of polyps, such as obstruction, gastrointestinal bleeding or intussusception. The latter is the main cause of death in children (5). In addition to the mentioned neoplasias, patients with Peutz-Jeghers are at risk of developing neoplasms of ovary, fallopian tubes, testis, thyroid, lung, gallbladder and bile ducts (6).

The current imaging modalities used for diagnosis and follow-up of the Peutz-Jeghers syndrome count with a variety of options that, from less to more complex, Include conventional baritone studies, axial studies such as Computed Tomography (CT) or MRI (with or without enteroclisis) and videoendoscopy with capsule (VEC) or with double ball (1).

Axial studies have replaced in recent years the barium studies as initial evaluation because of their high spatial resolution, its ability to evaluate the intestine without interposed loops, the possibility of evaluating extraintestinal structures and accessibility in the current medium (7). These studies can be with or without enteroclisis; the use of this has the advantage of a greater intestinal distention and, therefore, a more complete evaluation. However, in addition to its higher cost, it carries the risk of ionizing radiation in the case of CT, and the risks involved in the use of an invasive technique (7).

Although the diagnostic performance of CT with respect to MRI is similar for the study of this disease, there are advantages and disadvantages when either of the two modes is selected: the CT offers greater spatial and temporal resolution, lower costs, greater accessibility and shorter acquisition times of images compared to MRI. On the other hand, MRI is superior in detection of extraenteric abnormalities, allows an excellent differentiation with high contrast in the soft tissues and does not use ionizing radiation, which allows to repeat the study as often as necessary for the diagnosis and follow-up of these patients without accumulating doses of radiation.

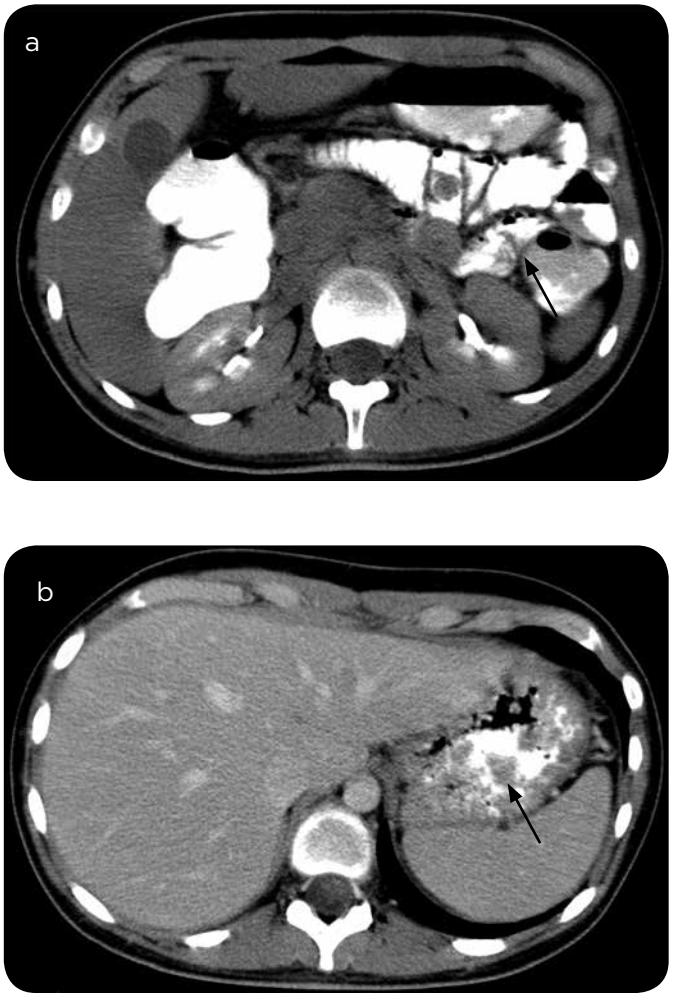

Figure 1. (a) Polyps that compromise the jejunum and (b) the gastric chamber.

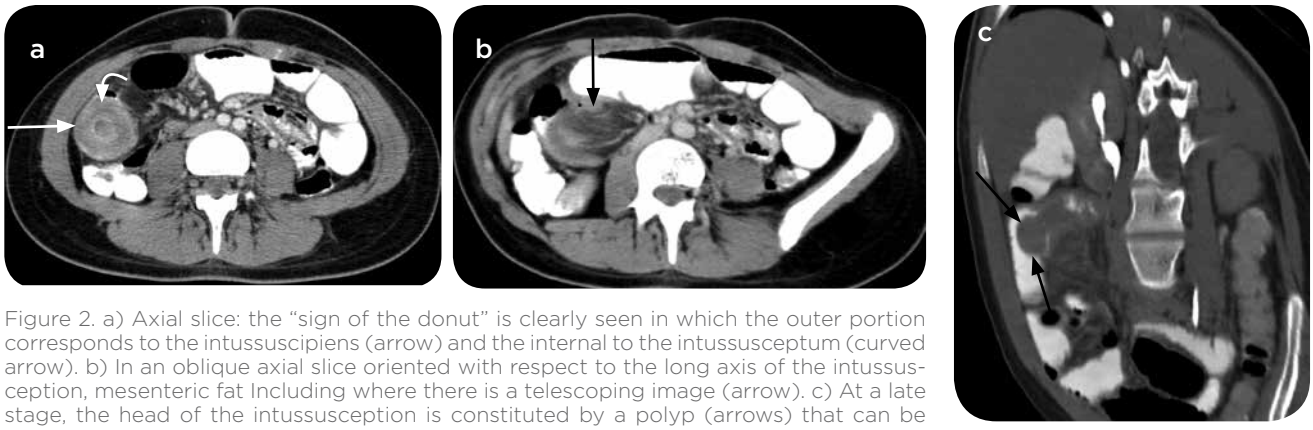

Figure 2. a) Axial slice: the "sign of the donut" is clearly seen in which the outer portion corresponds to the intussuscipiens (arrow) and the internal to the intussusceptum (curved arrow). b) In an oblique axial slice oriented with respect to the long axis of the intussusception, mesenteric fat Including where there is a telescoping image (arrow). c) At a late stage, the head of the intussusception is constituted by a polyp (arrows) that can be defined clearly in oblique sagittal reconstruction.

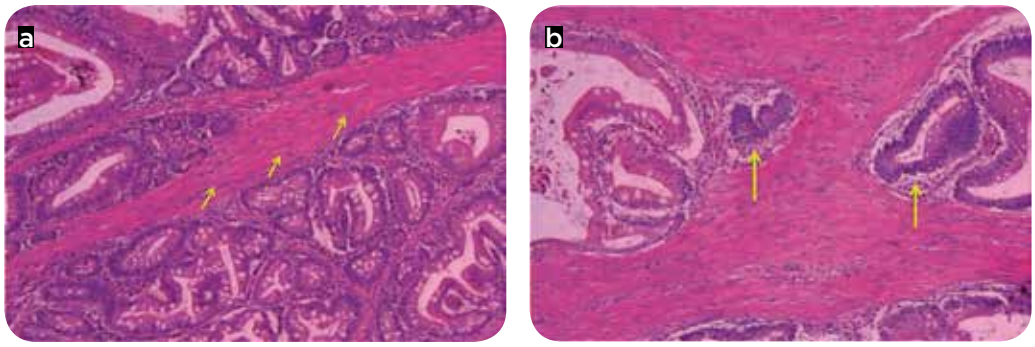

Figure 3. a) Polyp of Peutz-Jegher of the small intestine. The muscle of the thickened mucosa and arboriform or branched (arrows) is observed, covered by thin intestinal mucosa. b) Peutz-Jegher small intestine polyp with pseudoinvasion characterized by benign mucinous cysts through the intestinal wall (arrows).

Based on the above, the most recent studies recommend the evaluation of small bowel pathologies with MR as first line, as long as it is available and adjusted to the condition of the patient (7,8).

The main findings in the axial modalities (CT, MRI) in this syndrome are the hamartomatous polyps, which can compromise the entire alimentary tract, except the esophagus, with the jejunum and ileum as the most commonly affected, followed by the duodenum, colon and stomach (4,8). Polyps can vary in their size from a few millimeters to several centimeters and can be sessile or pedicled. The distribution of these lesions may be clusters or separated by healthy mucosa. It must be taken into account that these findings may be found in other entities such as polyposis adenomatous polyps (adenomatous polyps, premalignant [3]), juvenile polyposis (adenomatous polyps with degeneration to adenomas) and, subsequently, carcinoma [3]), Cowden's syndrome (hamartomatous polyp, sessile, non-premalignant [4]) and Cronkhite-Canada (sessile, hamartomatous polyps with occasional malignant degeneration [3]). In some patients the polyps may be unique or very few and, in these cases, may be confused with hairy adenomas or adenocarcinomas (9).

Intussusception is a frequent complication in patients with Peutz-Jeghers syndrome and usually occurs in the small intestine. It can be observed in different modalities of image: by ultrasound one can identify the 'sign of the doughnut' when obtaining images of the short axis of the compromised segment or it can be seen as a pseudokidney when obtaining longitudinal images. In tomography it is revealed as a complex soft tissue mass when the intussusceptum handle is centrally observed and the intussuscipiens handle peripherally. Low density portions corresponding to mesenteric fat associated with mesenteric vessels accompanying the intussusceptum can be seen. In the images in which the X-ray beams are perpendicular to the short axis of the intussusception, there is a 'target sign' and in the images parallel to the long axis it is observed as a configuration in 'sausage' (1,10) (Figure 2).

Patients with this condition require strict follow-up due to the already described risk of malignancy. Currently, it is recommended that specific screening tests for breast, testis, ovaries, pancreas and pelvis with ultrasound, every 2 years, and yearly mammography scans be made from the age of 25. Evaluation of the gastrointestinal tract may be carried out with endoscopy and intestinal transit every 2 years and colonoscopy every 3 years (3,10,11). MRI enterography is an alternative to the endoscopic capsule for patient surveillance with large polyps because it is more accurate for the location and estimation of the size of these anomalies.

On the other hand, there have not been found any significant differences between these two techniques for the detection of polyps greater than 10-15 mm, which are the most relevant for patients with this syndrome because they involve a greater risk of complications and malignant degeneration (7,12). It must be taken into account that the TruFISP and VIBE sequences with fat suppression are the most useful for the detection of polyps. Polyps are identified as defects of fillings of low signal in TruFISP sequences and typically show a similar enhancement to that of the intestinal wall after administration of gadolinium (12,13).

Discussion

This case is relevant because Peutz-Jeghers syndrome is a rare entity that occurs from 1:8300 patients up to 1:200,000; However,

clinicians and radiologists should take it into account since the identification and follow-up of these patients can decrease morbidity and expenses to the health system, mainly, because of the high risk of developing malignancies (14). In the case studied, typical clinical and tomographic signs facilitated the diagnosis, such as hyperpigmentation of the skin and predominance of polyps in the small intestine, in addition, of the known history of the disease inherited by paternal line in concordance with its autosomal dominant mode of transmission (1). The patient manifested abdominal pain, so that ileoileal intussusception was diagnosed, which represents one of the 3 most frequent complications in these patients (3). The majority of patients with Peutz-Jeghers with intussusception are given expectant management, since a large proportion of these are reduced spontaneously.

Conclusion

Peutz-Jeghers disease is a rare autosomal dominant entity, characterized by the presence of hamartomatosous polyps in the gastrointestinal tract and hyperpigmented lesions on the skin and mucous membranes associated with a high risk of developing neoplasms in different organs, as well as complications associated with the presence of intestinal polyps such as intussusception. Images play an important role in the diagnosis of the entity, detection of complications and long-term follow-up of affected patients, therefore every radiologist must know and be able to identify it.

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