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Case Report

Intestinal intussusception secondary to polyposis in Peutz-Jeghers syndrome

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ABSTRACT

Peutz-Jeghers syndrome (PJS) is an autosomal dominant syndrome characterized by multiple hamartomatous polyps in the gastrointestinal tract, mucocutaneous pigmentation. We present a case of intestinal intussusception secondary to these polyps.

Keywords: Peutz-Jeghers syndrome, Polyps, Intussusception, Intestinal occlusion

INTRODUCTION

The relationship between mucocutaneous pigmentation and intestinal polyposis was first reported by Peutz in 1921, and it was not until 1949 that Jeghers et al wrote the definitive clinical description of Peutz-Jeghers syndrome (PJS) in detail.[1]

PJS is an autosomal dominant syndrome characterized by multiple hamartomatous polyps in the gastrointestinal tract, mucocutaneous pigmentation, and an increased risk of gastrointestinal and non-gastrointestinal cancer.[2]

Other causes of intestinal polyps include familial adenomatous polyposis, which can be divided into attenuated <100 adenomas which is associated with a later onset of colorectal cancer and the absolute risk is thought to be lower than in those with a classic phenotype (>100 adenomas).[3]

Polyps are hamartomas that characteristically contain a smooth muscle proliferation that extends into the lamina propria in an arborizing fashion; the overlying epithelium is normal. Estimated prevalence of 1:8,000 to 1:200,000 births. In Japan the estimated prevalence is approximately 50,000 to 200,000 live births. Approximately 17% to 50% of cases are solitary cases with no family history of the disease.[2,4]

The syndrome can occur in any ethnic group, and affects men and women equally. Patients with PJS have an increased risk of developing malignancies; the average age of cancer development is 42 years.[5]

Gastrointestinal polyps develop in the first decade of life, and most patients present with symptoms between the ages of 10 and 30 years. Ten to 20 percent of individuals with PJS have no family history and are presumed to have PJS due to de novo mutations.[2]

PJS is most often due to germline mutations in the STK11 (LKB1) gene encoding a serine threonine kinase mapped to chromosome 19p13.3, a tumor suppressor gene that regulates the activity of members of the AMP-activated protein kinase (AMPK) family to control multiple cellular processes including cell polarity, metabolism, and

apoptosis. There are several polyposis-associated genes including PTEN, GREM1, POLE/POLD1, and biallelic NTHL1. The predominant clinical feature of PJS is gastrointestinal polyposis where it can cause bleeding, anemia, and intussusception.[2,3]

Mucocutaneous pigmented macules (melanin spots) are present in more than 95%, caused by pigment-laden macrophages in the dermis, they are flat, bluish-gray to brown spots, 1 to 5 mm in size. Gastrointestinal hamartomatous polyps are present in the majority of patients. They occur most frequently in the small intestine (60% to 90%) and more specifically in the jejunum, they can be found throughout the gastrointestinal tract, including the stomach (15% to 30%) and colon (50% to 64%).[2]

The cumulative risk of colorectal cancer is 5% in the general population with 39% in patients with PJS, with an average diagnostic age of 42-46 years. In the stomach, it is 29% for patients with PJS with an average age of diagnosis of 30-40 years. In the small intestine, it can be up to 13% with an average age of diagnosis of 37-42 years. Other related types of cancer: breast, uterus, ovary, testicular and pancreas, from 11% to 36%.[2,3]

Related complications include: intestinal obstruction, bleeding, mesenteric ischemia, gastric outlet obstruction syndrome and anemia. Obstruction caused by intussusception or occlusion of the gastrointestinal lumen by the polyp occurs in up to 69% of patients, most frequently in the small intestine.[2]

Intussusception has been seen at the age of 10 years in 33% of cases and at the age of 20 years in 50%. The risk increases with polyps >15 mm. In 1991, Chuang and Chen reported the first pediatric case of duodenal-jejunal intussusception secondary to hamartomatous polyps of Brunner's glands in the second portion of the duodenum without involving the ampulla of Vater.[6,7]

PJS is based on the clinical findings of having at least 2 of the 3 clinical criteria listed for a positive diagnosis: family history; multiple dark blue to brown pigmented lesions (macules) on the mucous membranes and skin, most often intraoral on the buccal mucosa or gingiva, lips, perioral, fingertips, palms, and soles; and hamartomatous intestinal polyps.[5]

Clinical diagnosis with the presence of any of the following: two or more Peutz-Jeghers-type hamartomatous polyps of the gastrointestinal tract; characteristic mucocutaneous hyperpigmentation of the mouth, lips, nose, eyes, genitals, or fingers in a person with a family history of PJS; any number of Peutz-Jeghers polyps in an individual with a family history of PJS in a first-degree relative; and any number of Peutz-Jeghers polyps in a person with the characteristic mucocutaneous pigmentation of PJS.

Individuals who meet the clinical criteria should undergo genetic testing to detect germline mutation in the STK11 gene, however, the absence of this does not exclude the diagnosis since not all associated mutations have been identified. Currently, magnetic resonance enterography (MRE) and video capsule endoscopy (VCE) are the most commonly used imaging modalities for the detection of small bowel polyps.[2,3]

VCE allows for better visualization of small bowel polyps than barium radiographs and is recommended as a first-line surveillance procedure. In children, the capsule can be deployed into the duodenum after upper endoscopy. Magnetic resonance enterography (MRE) is a reliable procedure for the detection of larger small bowel polyps with sensitivity similar to VCE and avoids radiation exposure from CT enterography. Other alternatives include MRI and CT, but these are poorly tolerated.[6]

At endoscopy, polyps may be sessile, pedunculated, or lobulated. The number of polyps varies from 1 to more than 20 per segment of bowel, although some patients have solitary lesions. Polyp size ranges from 0.1 to more than 5 cm in diameter. ESGE recommends baseline esophagogastroduodenoscopy and colonoscopy at age 8 years in asymptomatic individuals with PJS.[2,3]

Histologically, PJS polyps are hamartomas that characteristically contain a smooth muscle proliferation extending into the lamina propria in an arborizing fashion; the overlying epithelium is normal. Epithelial malpositioning involving all layers of the bowel has been reported in approximately 10 percent of small bowel polyps in PJS. Epithelial malpositioning, possibly due to mechanical forces, may extend into the serosa and be misdiagnosed as a well-differentiated adenocarcinoma.[2]

PJS, Bannayan-Riley Ruvalcaba syndrome (BRRS) and Cowden syndrome belong to a family of hamartomatous polyposis syndromes. They therefore share similar clinical features, like gastrointestinal polyposis, but differ in the mutation of genetic loci.[5]

Pigmented intraoral mucocutaneous macules can be found in Laugier-Hunziker syndrome (LHS), but appear later in life after childhood. Laugier-Hunziker syndrome is not associated with gastrointestinal polyposis.[5,8]

Duodenal adenomatosis is the most frequent extracolonic manifestation in familial adenomatous polyposis and therefore implies an important differential diagnosis.[3]

Another differential diagnosis of causes of intussusception is Meckel's diverticulum which is the most common pathologic site in most case series in children, followed by polyps.[8]

Endoscopic polypectomy should be performed for polyps >0.5 cm detected during upper endoscopy and colonoscopy. All other small bowel polyps that are

symptomatic or >1 cm in size should be resected to reduce the risk of related complications.[2]

Balloon-assisted enteroscopy (BAE) can remove distal small bowel polyps with or without laparotomy. The efficacy and safety of BAE for the treatment of small bowel polyps in PJS has been established in children with PJS. BAE and polypectomy decrease the need for intraoperative enteroscopy and enterotomy, which should be reserved for individuals affected with large, distal small bowel polyps beyond the reach of BAE.[6]

CASE REPORT

6-year-old female, diagnosed with PJS, with a history of endoscopic gastric and colonic polypectomy in 2022, with a histopathological report of hyperplasia. The current condition begins in June 2023 with intestinal occlusion, which remits with conservative management. After a month, she restarted with intolerance to the oral route, fever and biliary vomiting, denying symptoms suggestive of gastrointestinal bleeding. On physical examination, she presented increased peristalsis, generalized hypertympanism, without peritoneal irritation. An X-ray showed distention of the loops (Figure 1), and an abdominal ultrasound reported intestinal intussusception; all of this associated with a leukocytosis of 26×10^3/ul and persistence of the systemic inflammatory response syndrome, for which surgical management was offered. A laparotomy with a transverse supraumbilical incision was performed, and the following findings were found: 2×1 cm polyps at 10 and 30 cm, 3×2 cm at 50 cm, and 4×3 cm at 180 cm from the fixed loop (Figure 2), causing segmental intussusception in the small intestine (Figure 3); the last segment described was not reducible to taxis. A 10 cm segment was resected at 170 cm from the fixed loop, and a manual end-to-end anastomosis was made in two planes; to resect the other polyps, a longitudinal enterotomy of 5 cm was performed on the antimesenteric border at 30 cm from the fixed loop (Figure 4), and polypectomy of the 3 adjacent polyps through this enterotomy (Figure 5), with closure of the polypectomy mucosa with Lembert stitches and transverse enterorrhaphy in two planes.

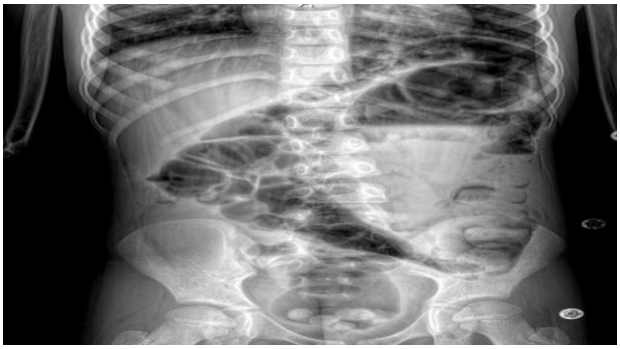

Figure 1: Abdominal X-ray: intestinal occlusion.

An open drain was left directed to the pelvic cavity and anastomosis site. During the postoperative period, the patient was fasted for 72 hours and a polymeric diet was started, with no incidents. The decision was made to discharge the patient on the 6th postoperative day. Currently, after one year of follow-up, there has been no recurrence of intestinal occlusion, with pathological reports of benign hamartomatous polyps (Figure 6).

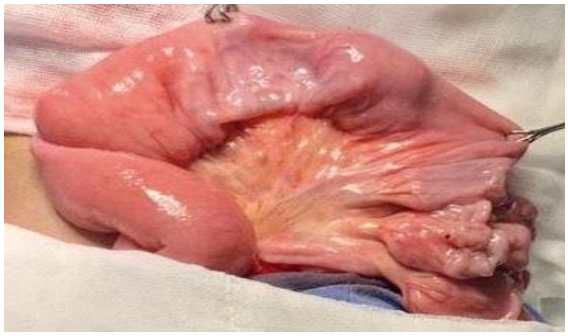

Figure 2: Intestinal polyp.

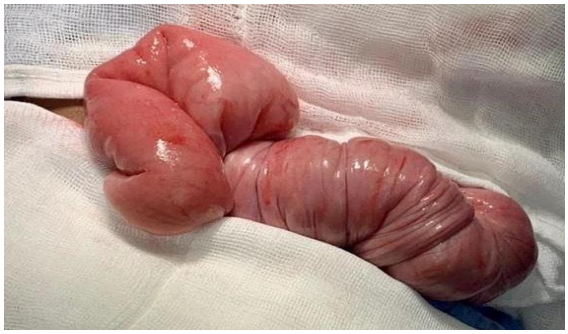

Figure 3: Intestinal intussusception.

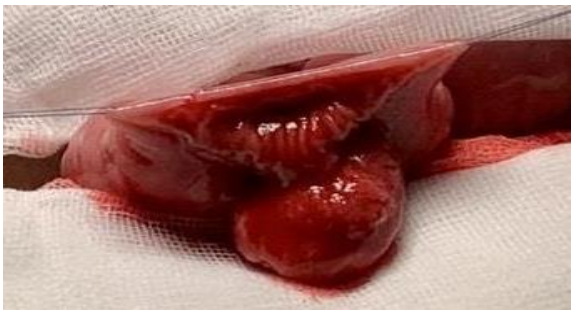

Figure 4: Exteriorization of polyp by enterotomy.

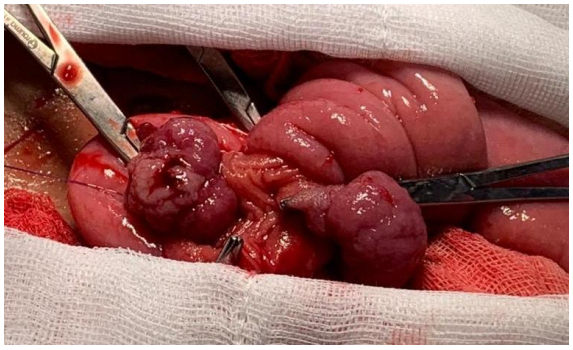

Figure 5: Polypectomy.

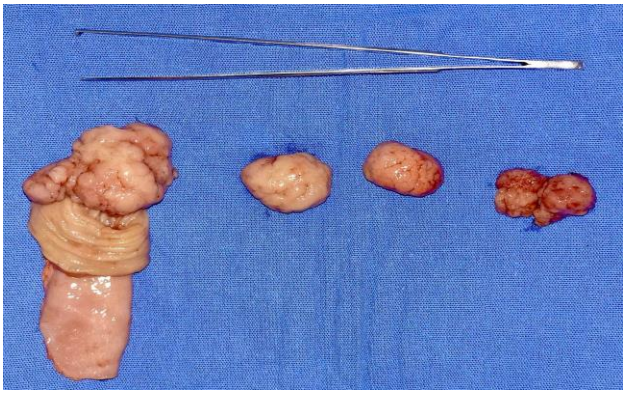

Figure 6: Intestinal polyps.

DISCUSSION

In terms of treatment, guidelines published by the Japan Gastroenterological Endoscopy Society (JGES) and the European Society of Gastrointestinal Endoscopy (ESGE) recommend polypectomy by BAE for small bowel polyps larger than 10 to 15 mm. Polyps larger than 10 to 15 mm, symptomatic polyps, and rapidly growing polyps should be resected. BAE can usually be performed in patients aged 3 years and weighing 14 kg or more.

Acute pancreatitis and post-polypectomy syndrome have been reported as complications. Ischemic polypectomy using a detachable snare or clip has been described and can treat many polyps in a shorter period than endoscopic resection and is less likely to cause complications. When polyps are exceptionally large and difficult to resect or cannot be reached by BAE, intraoperative endoscopic polypectomy or removal through a bowel incision should be considered.[4]

Indications for surgery include failure to achieve endoscopic control of polyps, either due to the size or number of polyps and the presence of neoplasia. Surgery may also be necessary in patients with small bowel obstruction or intussusception. Most duodenal tumors causing duodenojejunal intussusception are benign. Reduction of the duodenojejunal invagination and local excision of the tumor is sufficient.[2,7]

Wagner et al mention that surgical reduction of intussusception should be performed without delay to avoid necrosis and bowel resection. When ischemia is reversible, polypectomy alone should be performed. In our case report, the segment 170 cm from the fixed loop was not found to be reducible or viable, we decided to resect the affected segment and opted for transintestinal polypectomy of the jejunal polyps to avoid increasing surgical morbidity.

In addition, intraoperative enteroscopy via enterotomy is recommended to find and remove polyps larger than 15 mm. If enteroscopy is not available, illumination and thorough palpation of the small intestine is recommended to palpate and remove larger polyps.[9]

Patients with PJS may develop gastrointestinal polyposis as early as age 10. Therefore, it is imperative to evaluate the small intestine with upper endoscopy in addition to colonoscopy beginning in early adolescence.[5]

CONCLUSION

Intussusception is one of the causes of intestinal occlusion in patients with polyps. Endoscopic polypectomy is the technique of choice; however, polypectomy with exteriorization of the polyps by enterotomy is an adequate option since minimally invasive procedures are not available. With this, we avoid intestinal resection in a condition with a high recurrence of polyps such as PJS.

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REFERENCES

1. McGarrity TJ, Amos CI, Baker MJ. Peutz-Jeghers Syndrome. GeneReviews®. University of Washington, Seattle. 2021.
2. Daniel C, Chung M. Peutz-Jeghers syndrome. MedlinePlus. 2023. Available at: https://medlineplus-gov.translate.goog/spanish/ency/article/000244.htm?_x_tr_sl=es&_x_tr_tl=en&_x_ tr_hl=en&_x_tr_pto=sc. Accessed on 09 June 2024.
3. van Leerdam ME, Roos VH, van Hooft JE, Dekker E, Jover R, Kaminski MF, et al. Endoscopic management of polyposis syndromes: European Society of Gastrointestinal Endoscopy (ESGE) Guideline. Endoscopy. 2019;51(9):877-95.
4. Yamamoto H, Sakamoto H, Kumagai H, Abe T, Ishiguro S, Uchida K, et al. Clinical Guidelines for Diagnosis and Management of Peutz-Jeghers Syndrome in Children and Adults. Digestion. 2023;104(5):335-47.
5. Bohórquez EJF, Rincón DSQ, Gamboa YVC, Forero MCA. Peutz-Jeghers syndrome: case report. Rev Colomb Gastroenterol. 2022;37(4):502-6.
6. McGarrity TJ, Amos CI, Baker MJ. Peutz-Jeghers Syndrome. GeneReviews®. University of Washington, Seattle. 2021.
7. Hwang CS, Chu CC, Chen KC, Chen A. Duodenojejunal intussusception secondary to hamartomatous polyps of duodenum surrounding the ampulla of Vater. J Pediatr Surg. 2001;36(7):1073-5.
8. Salazar, JH. Intussusception in children. UpToDate. 2024. Available at: https://www.uptodate.com/contents/intussusception-in-children?search=intususcepci%C3%B3n%20en%20ni%C3%B1os&source=search_result&selectedTitle=1%7E122&usage_type=default&display_rank=1#H1. Accessed on 09 June 2024.

9. Wagner A, Aretz S, Auranen A, Bruno MJ, Cavestro GM, Crosbie EJ, et al. The Management of Peutz-Jeghers Syndrome: European Hereditary Tumour Group (EHTG) Guideline. J Clin Med. 2021;10(3):473.

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