CASE REPORT

Giant rectal polyp prolapse in an adult patient with the Peutz-Jeghers syndrome

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SUMMARY

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant intestinal polyposis syndrome characterised by the presence of hamartomatous polyps and mucocutaneous pigmentation. Prolapse of the polyps through the anus is an infrequent manifestation in children with PJS, and this complication is extremely rare in adult patients. We report the case of a 30-year-old man recently diagnosed with PJS who was seen at the emergency department because of the abrupt onset of severe anal pain with a foreign body sensation in the anal canal and rectal bleeding.

Physical examination revealed a giant prolapsed polyp.

BACKGROUND

Peutz-Jeghers Syndrome (PJS) is an autosomal dominant intestinal polyposis syndrome characterised by the presence of hamartomatous polyps and mucocutaneous pigmentation.^{1 2} Its prevalence is estimated at one case per every 8300 to 29 000 live births.³ The major complications in these patients include intussusception of the small intestine and the increased risk of intestinal and extraintestinal malignancies.⁴ Patients often present with rectal bleeding, intestinal invagination, anaemia and mucocutaneous pigmentations in infancy or early childhood.⁵ In children, a polyp prolapsed through the anal canal is an unusual manifestation of this syndrome,⁶ and in adults this condition is extremely rare.

CASE PRESENTATION

A 30-year-old man with a history of chronic abdominal pain episodes, constipation and rectal bleeding was seen at our unit. Physical examination revealed the presence of hyperchromic spots on the lips and in the mucosal palate, with no other relevant findings. His father was diagnosed with PJS at the age of 35 years. A colonoscopy was performed, revealing the presence of multiple sessile polypoid lesions of different sizes from 3 mm to 5 cm along the entire colon and terminal ileum. Histopathology reported multilobulated polyps with a papillary surface and branching bands of smooth muscle covered by hyperplastic glandular mucosa, consistent with hamartomatous polyps. Thus, PJS diagnosis was established in our patient and a surveillance programme was offered. Upper gastrointestinal (GI) endoscopy and small bowel capsule endoscopy were performed, revealing

multiple hamartomatous polyps along the small bowel with no suspicion of gastrointestinal malignancies.

Three months later, the patient was seen at the emergency department because of the abrupt onset of severe anal pain with a foreign body sensation in the anal canal and rectal bleeding. Physical examination revealed a giant, reddish-blue prolapsed polyp with a necrotic aspect (figure 1).

INVESTIGATIONS

With the patient under sedation, digital manoeuvres were performed to reintroduce the prolapse. Colonoscopy was then performed and a hot-snare polypectomy using submucosal infiltration with epinephrine (1:10 dilution) was carried out (figure 2A–D). Macroscopically, a large 5×6 cm polyp was found. Histopathology reported multilobulated polyps with a papillary surface and branching bands of smooth muscle covered by hyperplastic glandular mucosa with recent haemorrhage (figure 3). At 2 weeks after the procedure, an anorectal manometry was performed (high-resolution 12-sensor probe, Given Imaging, Yoqneam, Israel) and no abnormalities were detected. An endoanal ultrasound was carried out and no sphincter defects were found.

DIFFERENTIAL DIAGNOSIS

Mucosal rectal prolapse.

TREATMENT

Medical treatment with psyllium was recommended to avoid constipation and the patient remains asymptomatic. He continues to undergo screening and surveillance protocol.

Figure 1 Giant polyp prolapsed through the anal canal.

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Figure 2 Endoscopic polypectomy. (A) Giant reddish-blue polyp with a necrotic aspect. (B–C) Hot snare polypectomy. (D) Polypectomy specimen.

OUTCOME AND FOLLOW-UP

The patient is currently asymptomatic and continues to have regular visits at our unit.

DISCUSSION

PJS is one of the hamartomatous polyposis syndromes, and is the result of the dominant inheritance of genetic mutations present in the 19p 13.3 and 16q chromosomes that encode a serine tyrosine kinase known as STK11/LKB1.^{7 8} According to the WHO criteria, PJS is based on: (1) the presence of three or more PJ polyps confirmed by histology; (2) any number of PJ polyps and a family history of PJS; (3) characteristic mucocutaneous pigmentation and a family history of PJ and (4) any number of PJ polyps and characteristic mucocutaneous pigmentation.^{8 9}

The histological hallmark of the polyps is the appearance of an arborescent stroma (tree-like appearance) as a result of a thick smooth muscle core derived from the muscularis mucosae

covered by a combination of glandular crypts and other mature cells such as goblet, Paneth and absorptive cells.^{3 7} The size of these polyps ranges from 0.5 to 5 cm and can be present along the entire gastrointestinal tract, but the small bowel is the most affected segment (up to 96%), followed by the colon and rectum (~25%).^{8 10}

Most of the patients develop GI symptoms during childhood or adolescence, such as chronic abdominal pain as a consequence of ulceration, bleeding or intussusception of larger polyps, usually in the small intestine.^{8–11} For example, in a case series of 11 patients (mean age of 6 years at symptom onset), 8 were operated on during a follow-up period of 15 years; 6 had at least 1 small bowel resection and 4 had repeat surgery for recurrent intussusceptions.¹²

Although our patient had a history of chronic abdominal pain, prolapse of mucosal and anal polyps is not a common condition in adult patients, and therefore we thought it would be of interest to report this case. This condition has been observed as the initial manifestation in children aged under 2 years, together with rectal prolapse (in up to 24% of cases).^{6 13} In one of the largest PJS series in the world (222 cases in Japan, mean age 23 years), only 7.2% had extrusion of polyps through the anus as a complication.

Multiple mechanisms are related to polyps that prolapse through the anus. It seems that this condition is seen more often in children because during the first years of life there is less fat in the ischioanal fossa, and so there is less pressure provided for this functional component of the perineum.^{6 13 14} Increased straining during bowel movements triggered by diarrhoea, a frequent condition at this age, may also play a role.^{6 13} In adults, the main predisposing factors for prolapse include defects or dysfunction of the anal sphincter or conditions that induce increased intra-abdominal pressure, such as chronic constipation.¹⁵ In our case, the patient had no obvious alterations in the integrity of the anal sphincter that predispose to prolapse, but he suffered from constipation.

Although there is no consensus as to how to treat prolapsed polyps in PJS, the options include conservative treatment, endoscopic resection or even ultralow anterior resection.¹⁶ In our case, the hot-snare polypectomy was successful.

Figure 3 Histopathology analysis of polyps stained with H&E. (A) Photomicrograph of a Peutz-Jeghers (PJ) polyp with loss of continuity of the surface epithelium, sheets and fibrin neoformation vessels (H&E, $\times 5$); (B) PJ polyp with severe bleeding in the lamina propria and loss of surface epithelium (H&E, $\times 5$); (C) Smooth muscle bundles in the lamina propria and glandular crypts, the characteristic morphological hallmark for PJ polyps, are shown as well as their adenomatous aspect (H&E, $\times 10$); (D) This image clearly shows that the crypt epithelium consists of absorptive, goblet and Paneth cells (H&E, $\times 40$).

Since patients with PJS have a well-established increased risk of malignancies and many die at a relatively young age, screening and surveillance programmes are recommended.¹¹

Learning points

- Peutz-Jeghers syndrome (PJS) is an autosomal dominant intestinal polyposis syndrome characterised by the presence of hamartomatous polyps and mucocutaneous pigmentation.
- Prolapse of the polyps through the anus is an infrequent manifestation in children with PJS, and this complication is extremely rare in adult patients.
- The main predisposing factors for prolapse include defects or dysfunction of the anal sphincter or conditions that induce increased intra-abdominal pressure.

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Competing interests None declared.

Patient consent Obtained.

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