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Peutz-Jeghers Syndrome: A Case Report

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Abstract **Case Report**

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by hamartomatous gastrointestinal polyposis and mucocutaneous pigmentation. We report here the case of a 28-year-old young woman presenting with PJS revealed by an acute small bowel obstruction at the MED VI University Hospital of Marrakech. Surgical exploration revealed hamartomatous polyps necessitating segmental resection. Anatomopathological examination confirmed the diagnosis. Postoperative recovery was uneventful. Surveillance revealed no abnormalities.

Keywords: Peutz-Jeghers syndrome, hamartomatous polyps, mucocutaneous pigmentation, small bowel obstruction, surgical resection.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant inherited disorder characterized by a combination of hamartomatous digestive polyposis and a predominantly periorificial mucocutaneous lentiginosis. Although uncommon, this syndrome can lead to serious, mechanical complications such as intestinal obstruction and bleeding. It is sometimes associated with malignant, digestive or extra-digestive tumors. We report here the case of a 28-year-old young woman presenting with PJS revealed by an acute small bowel obstruction at the MED VI University Hospital of Marrakech.

CASE PRESENTATION

This concerns Miss L. H., aged 28, single and with a history of unexplored episodes of spontaneously resolving intermittent abdominal pain, and a history of small black spots on her lips around the age of 4 which gradually disappeared until the age of 24, with no other particular pathological history, particularly personal or family. She was admitted to the emergency department with an acute abdomen characterized by severe abdominal pain, vomiting and mechanical bowl obstruction. Physical examination revealed a conscious patient with blood pressure of 100/70 mmHg, heart rate of 90 beats per minute and respiratory rate of 17 cycles per minute, with notable abdominal distension and diffuse tenderness. The abdominal CT scan revealed a small bowel volvulus with signs of intestinal distress, and an emergency surgery was performed.

During the exploratory laparotomy, as soon as the peritoneum was opened, a sero-hematic effusion was detected, and upon exploration, an ileo-ileal intussusception with intestinal distress was observed. After de-intussception, two pedunculated intraluminal lesions were discovered, one 20 cm from the angle of Treiz measuring 5.5 cm x 4 cm with an implantation base of 3 cm and the other 30 cm from the angle of Treiz measuring 5 cm x 3.5 cm with an implantation base of 2 cm and a third small intestine tumor 1m 20 cm from the angle of Treiz, measuring 3 x3 cm with an implantation base of 1.5 cm, the procedure consisted of a segmental resection removing the three tumors with end-to-end ileo-ileal anastomosis. The anatomopathological study confirmed the presence of Peutz-Jeghers hamartomatous polyps, remodeled and focally invaginated with healthy resection margins and no histological signs of malignancy on three pieces of small intestine excisions. The postoperative follow-up was simple.

The diagnosis of Peuz-Jeghers syndrome was made based on the association of these hamartomatous polyps and the presence of lentiginosis on the patient's lips during childhood. The patient was well informed about the Peuz-Jeghers syndrome, the risk of recurrence of complications, the risk of cancer at a very young age, and the periodic check-ups that she must undergo.

The patient subsequently underwent a thourough clinical examination as part of the follow-up, an MR enterography that revealed no abnormalities, an esophagogastroduodenal fibroscopy and a colonoscopy, both of which were normal. She was then referred for gynecological examination as part of breast cancer screening but she has not shown up for any follow-up appointments since then.

DISCUSSION

Peutz-Jeghers syndrome is a rare but not exceptional disease. The global frequency is estimated at 1/200,000 births but it differs between each study; in the United States it is estimated at 1/60,000 to 1/300,000 births. It is a hereditary pathology with autosomal dominant transmission. Both genders are equally affected. It can occur in any racial or ethnic group [1, 2].

Peuz-Jeghers syndrome is characterized by the association of hamartomatous gastrointestinal polyps and mucocutaneous hyperpigmentation. Polyposis develops throughout the digestive tract, with the exception of the mouth. Histologically, the polyps are hamartomatous in nature corresponding to polyps of tubulovillous architecture without dysplasia. The axis of the villi is characterized by bundles of smooth muscle fibers, which differentiates them from adenomas. The tubes are covered with large cylindrical cells, at times enterocytic, at others goblet-shaped, with increased mucosecretion. Generally, these polyps do not have malignant potential [2].

In our case, the syndrome manifested itself at the age of 28 by an occlusive syndrome. The polyps were discovered during surgical exploration. The anatomo-pathological examination confirms that it was a hamartomatous polyp. Periorificial lentiginosis, or mucocutaneous hyperpigmentation, is rarely present at birth. In the literature, it appears before the fifth year of life then gradually decreases in adulthood and sometimes disappears completely. It is most pronounced at puberty. It results from cutaneous and/or mucous deposits of melanin, forming hyper-pigmented dark brown or black macules approximately 1 to 5 mm in diameter. It is almost constant but not pathognomonic, since a similar but often less spectacular appearance can be observed in certain normal subjects; it mainly affects the edges of the oral cavity (94%), eyes and nostrils (66%), hands and feet (62%); the perianal mucosa is much less frequently affected [3]. It usually precedes the appearance of polyps. In our patient, periorificial lentiginosis affected his lips. It was present in her childhood and disappeared before she reached adulthood. The diagnosis of Peuz-Jeghers syndrome was made based on the association of peri-orificial lentiginosis and hamartomatous polyps of the small intestine as well as her age. Peutz-Jeghers syndrome is characterized by its high risk of cancer, both digestive and extra-digestive. According to Lim et al, in a study of 240 patients with PJS, the risk of developing cancer was 1% before the age of 20; 19% before the age of 40; 63% before the age of 60 and 81% before the age of 70 [4]. The most common cancers in this analysis were digestive cancers. For extra-digestive cancers, breast cancer was the most common [5]. Lim et al reported that 8% of women with PJS developed breast cancer before the age of 40; 32% before the age of 60 [4]. In our case, as part of surveillance, we performed an Entero-MRI, esophago-gastro-duodenal fibroscopy and colonoscopy with no abnormalities and the patient was referred for a gynecological examination with possible echo-mammography but she was lost to follow-up. In Peuz-Jeghers syndrome, polypectomy is recommended for any polyp larger than 1 cm in diameter due to a risk of complications. Hemorrhage, intussusception or intestinal obstruction are clear indications for surgery. It should be borne in mind that complications can recur. Therefore, extensive intestinal resections,should be avoided as far as possible for they can can ultimately lead to short bowel syndrome[6]. This can be achieved through two techniques for resecting polyps in PJS: intraoperative enteroscopy and double-balloon enteroscopy [7].

The evolution of SPJ as well as the median survival of patients are difficult to specify. Generally, this syndrome is considered a benign condition, compatible with long-term survival under lifelong medical supervision, but evolution is threatened by the occurrence of acute intestinal obstruction, and sometimes recurrent hemorrhage, multiplying surgical interventions with their own risks and exposing patients to nutritional disorders secondary to short bowel syndrome [7]. Therefore, patients suffering from Puitz-Jeghers disease deserve special monitoring. Early detection of polyps is essential through periodic endoscopic examination. Every two to three years, patients should undergo abdominal-pelvic and prostatic ultrasounds, complete upper and lower gastrointestinal endoscopies and an enteroscanner. In our case, we carried out the first oesophagogastroduodenoscopy, colonoscopy as well as Entero-MRI all of which were normal, and we scheduled clinical monitoring for the patient every year, upper and lower fibroscopy every two years, but she has since been lost to follow-up.

CONCLUSION

Peutz-Jeghers syndrome is a rare condition predisposing to cancer. Our patient's case highlighted the importance of recognizing the clinical manifestations of this syndrome particularly in patients with a history of abdominal pain and anemia. A multidisciplinary approach involving gastroenterologists, surgeons and geneticists is essential to ensure optimal care of these patients. A monitoring protocol must be established for each patient with PJS throughout life to avoid complications and improve the quality of life of these patients.

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