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Case report

Combined endoscopic and surgical treatment for Peutz-Jeghers syndrome: a case report

G. Kouklakis[1], J. Kountouras[2], J. Moschos[1], E. Apostolakos[1], P. Zezos[1], C. Zavos[2], D. Chatzopoulos[2]

SUMMARY

We report a rare case of a 30-year-old man, with Peutz-Jeghers syndrome who was admitted to our clinic with recurrent occlusion episodes of the small intestine and underwent operations three times due to intestinal intussusception. The enteroscopic visualization of multiple sessile and peduncular polyps throughout the small intestine was the reason why combined surgical and endoscopic treatment was decided on to the remove the hamartomatous polyps from the intestine. A review of the relevant literature is cited.

INTRODUCTION

Peutz-Jeghers syndrome is expressed as intestinal hamartomatous polyposis in association with characteristic mucocutaneous pigmentation. It appears to be inherited as a single pleiotropic autosomal dominant gene with variable and incomplete penetrance.[1] Although the polyps of Peutz-Jeghers syndrome are not true neoplasms, they may increase in size progressively and cause obstruction of the small intestine or intussusception.[2] The polyps may be found in the stomach, small intestine or colon, but they tend to be most prominent in the small intestine.[2] Symptoms of acute upper gastrointestinal bleeding and chronic fecal blood loss may complicate this syndrome.[1]

[1]424 General Army Hospital, Thessaloniki, Greece, [2]Ippokration General Hospital, Second Medical Unit, Aristotle University of Thessaloniki, Greece

Author for correspondence:

Kouklakis Georgios MD PhD, 51 Androutsou street, 55132 Thessaloniki – Nea Krini, Greece, e-mail: gastro@hol.gr

Case report

We report a case of a 30-year-old man with Peutz-Jeghers syndrome, who was operated on three times for intestinal occlusion due to intestinal intussusception. These three operations – lysis of the intussusceptions, were performed once in a decade (no etiological treatment). The patient was admitted to our clinic with epigastric pain, nausea and vomiting due to the occlusion episode. He has characteristic pigmentetion of the P-J syndrome on the limbs and hands. There was negative family history for the syndrome.

The enteroscopic visualization with multiple sessile and peduncular polyps in the small intestine was the reason why the following combined surgical and enteroscopic intervention was decided on:

Surgical operation: Middle hyper-umbilicus laparotomy was performed. The opening of the abdomen revealed three intussusceptions at three different points of the small bowel, without occlusion. We also found multiple adhesions due to the previous operations. We carefully palpated the small intestine and localized the bigger polyps. The biggest was found 10-15cm beyond the ligament of Treitz. Here we performed enterotomy and removed a large polyp. Through enterotomy, we introduced the enteroscope and started to remove the polyps we found by snare polypectomy while the enteroscope was simultaneously handled by the surgeon. With this maneuver we removed several (n=53) polyps. Consequently, it was decided that a second and third enterotomy be performed in other portions, where large polyps were found and could not be removed with snare polypectomy. Through these latter points of enterotomy we also introduced the enteroscope once again, to remove polyps. A number of polyps remained in the intestinal lumen after the operation.

DISCUSSION

Peutz-Jeghers syndrome is a rare condition. The inheritance is autosomal dominant with variable and incomplete penetrance of the gene. In our patient the family history was negative for the syndrome. The occurrence in males and females is essentially equivalent.[3] There is strong relationship between the syndrome and genetic factors. Gene mapping studies have identified linkage to a locus on chromosome 19p[2,4] and other studies on chromosome 6.[2,5]

Peutz-Jeghers syndrome is characterized by:

1. Hamartomatous polyps throughout the gastrointestinal tract (most notably in the small intestine) that occur in 88% to 100% of individuals with Peutz-Jegher's syndrome. Polyps range in size from 0.1 to 3 cm in diameter and have a coarsely lobulated surface. Larger polyps are pedunculated, but smaller polyps are sessile.[2] In our patient there were many polyps and the larger were pedunculated. These polyps may become quite large, leading to bleeding presenting as acute upper gastrointestinal bleeding or chronic fecal blood loss, and to manifestations such as iron deficiency anemia,[2,5] intussusception or obstruction.[6]

2. Mucocutaneous pigmented macules (melanin spots) on the lips, buccal mucosa and skin.[6] It is a distinctive feature that is observed in more than 95% of affected persons. Lesions on the lips may fade after puberty, requiring the clinician to examine the buccal mucosa and other parts of the cutaneous surface.[3] The presence of this pigmentation should alert the clinician to this syndrome, but the skin lesions and intestinal lesions are occasionally inherited separately.[1] In our patient there was characteristic pigmentation on the lips and hands. Rare cases of malignant melanoma arising in hyperpigmented areas of the rectum and fingers have been reported.[2,7]

3. About 50% of these patients develop malignancies of the gastrointestinal tract (stomach, duodenum, jejunum, ileum and colon) and nonintestinal organs (breasts, gonads, pancreas).[6] Ovarian cysts and distinctive ovarian sex cord tumours are seen in 5% to 12% of female patients with Peutz-Jeghers syndrome.[1,8] Hormonally active Sertoli cell testicular tumours with feminizing features may occur in young boys with this syndrome.[1,9] Other cancers that may occur in this syndrome include pancreatic cancers, polyps or cancers of the biliary tree and gallbladder, and breast cancers which may be found in young women and may be bilateral.[1] In our case, a detailed laboratory and clinical examination did not reveal any gastrointestinal or extra-intestinal cancer.

The Peutz-Jeghers polyp is a unique hamartomatous lesion characterized by a grandular epithelium that is supported by an arborizing framework of well-developed smooth muscle that is contiguous with the muscularis mucosae.[1] The lamina propria is normal and the characteristic architecture of the lesion appears to derive chiefly from the abnormal smooth muscle tissue.[1] It is noteworthy that histological examination in our case was distinctive for this syndrome.

The average age of diagnosis in Peutz-Jeghers syndrome is 23 years in men and 26 in women.[1] Our patient was 30 year old. The diagnosis of this syndrome is based on careful examination of the cutaneous and muco-cutaneous regions for typical melanin spots and the evaluation of the gastrointestinal tract for polyps with upper and lower gastrointestinal endoscopy, enteroscopy (small bowel endoscopy) or small bowel radiography. Our patient had characteristic melanin spots on his lips and buccal surface and hands.

A rational therapeutic strategy for Peutz-Jeghers syndrome comprises endoscopic removal of any polyp larger than 5mm. On the other hand, surgical treatment is indicated for removal of any small bowel polyps that are symptomatic or larger than 1.5cm.[1]

Finally, combined endoscopic and surgical treatment which is performed with small bowel endoscopy and polypectomy during laparotomy,[1,10] achieves a clean intestine and allows longer asymptomatic periods.[1] Combine treatment is particularly important for those patients who will undergo repeated surgical interventions due to clinical manifestations while they are still young.[11] This combined treatment can avert multiple enterotomies and decrease bowel resection segments.[12,13] In our case, we performed only three enterotomies from which we removed the polyps and to date our patient is free of any overt intestinal occlusion symptoms or other symptoms due to Peutz- Jeghers syndrome.

After diagnosis has been established, some authors recommend the following for patients with Peutz-Jeghers syndrome: annual hemoglobin examination, annual ultrasonography of the pancreas, pelvis (women), and testes, upper and lower endoscopy per 2-years, small bowel X-ray every 2 years, mammography at the ages of 25, 30, 35, 38, thereafter per 2 years until the age of 50, then annually, and cervical smear every 3 years. The interval

between pancreatic ultrasonography and small bowel X-ray may be extended after several examinations yield negative results.[1] Our patient was informed about the examinations to follow and which include an annual hemoglobin examination, annual ultrasonography of the pancreas, upper and lower endoscopy and small bowel X-ray every two years and he agreed to follow this procedure. Two years after the procedure, the patient is in a very good condition and the follow up examinations that we recommended have produced no abnormal findings.

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