



PEUTZ-JEGHERS SYNDROME

Surgery

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ABSTRACT

Peutz-Jeghers syndrome is an autosomal-dominant hereditary cancer syndrome that carries a 39% lifetime risk of CRC characterized by benign hamartomatous, primarily GI, polyps, mucocutaneous pigmentation (dark blue or brown macules in the vermillion border of the lips, buccal mucosa, hands, and feet), and a high predisposition to many intestinal and extraintestinal cancers.

KEYWORDS

Peutz-Jeghers syndrome

INTRODUCTION

Peutz-Jeghers syndrome is an autosomal-dominant hereditary cancer syndrome caused by a mutation of the *STK11/LKB1* gene located on chromosome 19p. Approximately half of PJS cases are inherited from a parent; the remainder occur in patients with no family history and appear to result from a spontaneous mutation. Nearly 90% of Peutz-Jeghers Syndrome patients will develop hamartomatous polyps, most commonly in the small bowel, followed by the colon, stomach, and rectum in decreasing frequency. PJS patients have a 90% lifetime risk of cancer, including colorectal (most common), gastric, pancreatic, lung, breast, uterine, cervical, testicular, and ovarian.

Case Presentation

A 21-years-old male presented with complaint of pain abdomen, abdominal distension, vomiting and non-passage of stool and flatus for 1 day. History of ATT for 6 months, 10 years back for Cold abscess over right side of neck. On general physical examination perioral melanosis was present.



Figure 1- Small, Blue to Dark Brown Pigmented Macules on Lips

On per abdomen examination, abdomen was tense, distended, tenderness was present over whole abdomen. Ultrasound whole abdomen shows bowel loops in lumbar region which appear distended and showing telescoping of the proximal bowel loop into distal bowel loop suggestive of Intussusception. CECT abdomen showed clumping and twisting of small bowel loops with twisting of blood vessels and mesentery in the region of jejunum. There is dilatation of the proximal jejunal loop. Small segment telescoping of the twisted jejunal loops into the distal jejunum also noted-suggest closed loop small bowel obstruction due to Volvulus with short segment intussusception likely. Smaller area of twisting of small bowel loops, blood vessels and mesentery also noted in right iliac fossa region involving the ileal loops. Splenomegaly was present. Intra-operatively a 30 cm. intussusception bowel loop of jejunum and proximal ileum. 20 cm. intussusception bowel loop of ileum into ileum, 40cm. proximal to ileo-caecal junction. 10 cm. intussusception bowel loop of ileum into ileum, 20 cm. proximal to ileo-caecal junction. Multiple enlarged lymph nodes

throughout the intestine. Multiple polypoidal growth present at intussusception site as mentioned above.



Figure 2-6 Intraoperative Photographs

DISCUSSION

Peutz-Jeghers syndrome is an autosomal-dominant hereditary cancer syndrome characterized by benign hamartomatous polyp. Polyps differ histologically from juvenile polyps in that they arise due to an overgrowth of the muscularis mucosa rather than the lamina propria three or more histologically confirmed Peutz-Jeghers polyps; (2) any number of PJ polyps with a family history of PJS; (3) characteristic, prominent, mucocutaneous pigmentation with a family history of PJS; or (4) any number of Peutz-Jeghers polyps and characteristic prominent, mucocutaneous pigmentation. Screening begins at 8 to 10 years of age with an evaluation of the small bowel. If initial exam is normal, a repeat evaluation is recommended at the age of 18 and then at 2-to 3-year intervals. Males should undergo annual testicular physical examination starting at age 10 years, and females should undergo an annual pelvic examination and Papanicolaou stain starting at age 18 to 20 years. Women should have breast physical examinations every 6 months and yearly mammogram and breast MRI starting at age 25 years. Colonoscopy and upper endoscopy should start in the late teens and be repeated every 2 to 3 years for both genders. Pancreatic cancer screening involves endoscopic ultrasound or magnetic resonance cholangiopancreatography along with serum CA19-9 every 1 to 2 years starting at age 25 to 30 years. Polypectomy plays a key role in the management of PJS. Asymptomatic gastric or colonic polyps larger than 1 cm should be removed endoscopically. Small bowel polyps larger than 1 to 1.5 cm or those that are have grown rapidly should be removed to decrease future complications such as bleeding and intussusception. Surgery is most commonly reserved for symptoms, the most common being obstruction (caused by intussusception) and bleeding in the small bowel. The goal of surgery is to remove the affected segment, preserving as much bowel as possible. Intervention may require push enteroscopy or combined laparoscopy/ laparotomy with endoscopy in the operating room as these small bowel polyps may not be visualized by other means.

CONCLUSION

PJS patients require special surveillance that includes multiple organs, as it is associated with an increased risk of cancer in many organs (small bowel, stomach, pancreas, colon, oesophagus, ovary, lung, uterus, breast, testes, and others).

REFERENCES

1. Van Lier MG, Mathus-Vliegen EM, Wagner A, van Leerdam ME, Kuipers EJ. High cumulative risk of intussusception in patients with Peutz-Jeghers syndrome: time to update surveillance guidelines? *Am J Gastroenterol*. 2011 May;106(5):940-5.
2. Sherman S, Menon G, Krishnamurthy K. Peutz-Jeghers Syndrome. [Updated 2025 Feb 17]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan-. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK535357/>