



# Peutz-Jeghers Syndrome Causing Gastroduodenal Intussusception: A Rare Case Report

Gyanendra Chaudhary<sup>1</sup>, Sachin Agrawal<sup>2</sup>, Situ Situ<sup>3</sup>, Ananvay Dadu<sup>4</sup>

<sup>1</sup>Assistant Professor, Department of Surgery, Netaji Subhash Chandra Bose Subharti Medical College, Subharti Puram, NH-58, Delhi - Haridwar Bypass Road, Meerut, Uttar Pradesh - 250005, India.

<sup>2</sup>Associate Professor, Department of Radiology, Netaji Subhash Chandra Bose Subharti Medical College, Subharti Puram, NH-58, Delhi-Haridwar Bypass Road, Meerut, Uttar Pradesh - 250005, India.

<sup>3</sup>Assistant Professor, Department of Anaesthesia, Netaji Subhash Chandra Bose Subharti Medical College, Subharti Puram, NH-58, Delhi-Haridwar Bypass Road, Meerut, Uttar Pradesh - 250005, India

<sup>4</sup>Senior Resident, Department of Surgery, Netaji Subhash Chandra Bose Subharti Medical College, Subharti Puram, NH-58, Delhi-Haridwar Bypass Road, Meerut, Uttar Pradesh - 250005, India.

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## Corresponding Author

Gyanendra Chaudhary,  
Department of General Surgery,  
Netaji Subhash Chandra Bose Subharti  
Medical College,  
Subharti Puram, NH-58, Delhi-Haridwar  
Bypass Road, Meerut,  
Uttar Pradesh-250005, India  
Email: gyanendra.rims@gmail.com

## Abstract

Peutz-Jeghers syndrome (PJS) is a rare disease characterized by melanotic macular lesions over the pulp of fingers, in oral cavity mucosa, lips, and hamartomatous gastric and intestinal polyps. They usually present with intestinal intussusception or intestinal bleeding; gastroduodenal intussusception is an infrequent presentation. Endoscopic resection or complete surgical excision of the polyp is the treatment of choice. We here present a case of a seven year male with multiple gastric polyps presenting as gastric outlet obstruction.

## Introduction

Peutz-Jeghers syndrome (PJS) is a rare disease with an inheritance pattern of autosomal dominant. Its incidence is 1 / 8,300 to 1 / 2,80,000 and is characterized by melanotic macular lesions over the pulp of fingers, in oral cavity mucosa, lips, and hamartomatous gastric and intestinal polyps can be single to multiple polyps.<sup>1</sup> Exact etiology of this disorder is unknown but mutation in gene STK11 is found to be one reason.<sup>2</sup> Most common age of presentation is usually 30 to 40 years, but in about 33% of cases, they can be diagnosed before ten years. Usually, they present with intestinal intussusception or intestinal bleeding, and gastroduodenal intussusception is an infrequent presentation.<sup>3</sup> We present a case of a seven year male with multiple gastric polyps presenting as gastric outlet obstruction due to gastroduodenal intussusception.

## Case Report

A seven year boy was admitted with intermittent upper abdominal pain for one month and complaints of multiple episodes of non-bilious vomiting for two days. On examination, pallor, moderate dehydration and multiple melanotic lesions were present at the lower lip and pulp of fingers of both hands (Figure 1a). There was a lump palpable in the epigastric and right hypochondrium, with no sign of peritonitis. Ultrasound abdomen showed gastroduodenal obstruction but being so rare diagnosis,

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we decided to confirm it by computerized tomography (CT) scan, which confirmed ultrasound findings and showed the lead point as a polypoidal mass up to the duodenum (Figure 1b). Exploratory laparotomy with manual reduction of gastroduodenal intussusception was done. The stomach and bowel were healthy, and the stomach was intussuscepted from the body into the duodenum till the third part of the duodenum. Anterior wall gastrotomy was done which showed multiple polypoidal growths of the variable size found in the body and pylorus on the anterior wall and a few on the posterior wall of the stomach (Figure 2a, 2b). Through the gastrotomy site, intra-op endoscopy was done which showed normal bowel till ileum. All lesions of the stomach were resected along with the seromuscular layer (Figure 2c). The postoperative period was uneventful. Histopathology was consistent with features of Peutz-Jeghers Syndrome. The patient is in four months of follow-up and doing well.

## Discussion

PJS was first described by a British physician, Dr Connor in 1895 in twin sisters.<sup>4</sup> The gene which is involved in its pathogenesis is the STK11 gene or LKB1 gene. About 30 to 70% of cases of PJS have this genetic mutation.<sup>5</sup> These patients may have single to multiple polyps, with the most common site being small bowel followed by large bowel and stomach. They can be sessile or pedunculated or mixed, the pedunculated type is more common in the small bowel. The patients usually present between 30 and 40 years of age. However, around 30% cases present within 10 years of age, and rarely they can present in neonatal and infancy age groups also.<sup>3,6</sup>

Presentation of patients may be variable. They may have abdominal pain, melena, hematemesis, or can present with intestinal obstruction. They usually have melanin pigmentation over lips and pulp of fingers. Our patient, who presented with features of gastric outlet obstruction, also had melanotic pigmentation over lips and pulp of fingers. They are also associated with a high risk of breast, genital, or GIT malignancies.<sup>1</sup>

Initially, ultrasound (USG) and contrast studies were considered the investigation of choice but more advanced radiological investigations like computed tomography (CT) Scan or magnetic resonance imaging (MRI) has replaced conventional investigations. USG is still the initial investigation of choice, Doughnut or Target signs are typically seen in USG. Contrast-enhanced computed tomography

(CECT) Scans give information about underlying pathology as well as a complete assessment of the GIT system. Upper GI Endoscopy gives information on gastric and small bowel polyps. Wireless capsule endoscopy is getting popularity in early diagnosis, less invasiveness, and high yield.<sup>7</sup> Definitive treatment is required in case of complicating polyps. Endoscopy procedures like endoscopic polypectomy and double balloon endoscopic (DBE) excision of polyps are preferred over laparotomy. Laparoscopic reduction of intussusception is the preferred method for gastroduodenal or bowel intussusception. But in cases of peritonitis, malignancy, or intestinal obstruction, exploratory laparotomy is preferred. During laparotomy, endoscopy should be done through enterotomy to find out any missed polyps, as these are sources of recurrent intussusception. Complete excision of polyp along with mucosa, submucosa, and muscularis layer at base is advised to prevent recurrence. After the excision of the polyp, nearby mucosa is approximated for closure.<sup>8</sup> Routine screening is advised in cases of a diagnosed case of PJS or in children with a family history of PJS. This will help in the early detection of disease and the prevention of complications. If the patient is having clinical features of PJS, genetic screening for the STK11 gene should be done.<sup>9,10</sup>

## Conclusions

Peutz-Jeghers syndrome is less common in children and usually presents with hematemesis, melena, or recurrent intussusception. Gastroduodenal intussusception is a very rare complication causing gastric outlet obstruction. Early diagnosis is necessary to prevent complications and prevent laparotomy. Endoscopic excision of polyps is the treatment of choice. Routine follow-up is a must to prevent multiple laparotomies as this is a progressive disease.

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