



## Images in clinical practice

## Clinical criteria for Peutz-Jeghers syndrome diagnosis: Look for what is missing.

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Publication Data:  
Submitted: September 28,2020  
Accepted: October 31,2020  
Online: November 30,2020

This article was subject to full peer-review.



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### Abstract

The Peutz-Jeghers syndrome (PJS) is an autosomal dominant disorder characterized by hamartomatous gastrointestinal polyposis and mucocutaneous melanin pigmentation. In this report we discuss diagnostic circumstances of new PJS family.

**Keywords:**

Intestinal obstruction; intussusception; Peutz-Jeghers syndrome; polyps; surgery.

### Introduction

PJS is characterized by autosomal dominant inheritance. This syndrome is caused by mutations of the serine/threonine kinase 11 (STK11) tumor suppressor gene on the chromosome 19p13.3. Most of PJS complications are related to the evolution of the digestive polyps [1,2].

**Observations**

We report a case of a 14-year-old female with no personal or familial surgical history who presented for severe abdominal pain and recurrent vomiting in the last 24 hours. Her abdomen was distended tympanitic and tender. Investigation including abdomen X-rays erect and abdominopelvic ultrasound demonstrated acute intestinal obstruction due to a mass of the terminal ileum. The patient underwent a laparotomy. The findings were in favor of small bowel obstruction due to ileo-ileal intussusception. Resection of devitalized bowel and end to end anastomosis were performed (figure1). Postoperative course was uneventful. The gross examination of the specimen showed a 4 centimeters polyp. The histopathology confirmed its hamartomatous features. PJS was suspected. However, the investigations performed including gastroscopy, colonoscopy, and enteroclysis ruled out only one more small polyp of the left colon. The patient was assigned to regular follow up. One month later, the patient's 13-year-old brother presented to the emergency department for obstructive syndrome. Physical examination revealed pigmentation of the lips and the buccal mucosa (figure 2). This patient underwent a laparotomy. Ileo-ileal long intussusception on huge polyp was found. The polyp was resected via enterotomy after reduction (figure 3). There were no postoperative complications. The diagnosis of PJS was confirmed for both siblings. More investigations for all the family members as well as genetic study were indicated.

**Discussion**

Clinical diagnosis of Peutz-Jeghers syndrome is confirmed in the following situations: three or more histologically confirmed Peutz-Jeghers polyps, any number of Peutz-Jeghers polyps with family history of Peutz-Jeghers syndrome, characteristic mucocutaneous pigmentation with a family history of Peutz-Jeghers syndrome, any number of Peutz-Jeghers polyps and characteristic mucocutaneous pigmentation [3]. Small bowel intussusception in PJS is widely described in the literature [4]. Suspected PJS cases should undergo exhaustive screening for gastrointestinal polyposis. The capsule endoscopy seems to be more effective in the small bowel exploration [5]. Sporadic PJS exist but this diagnosis should be retained after clearance of the family members. In our report, the lack of some criteria did not allow the PJS diagnosis confirmation in our first patient. Precise clinical investigation could always rule out important diagnostic arguments such as the mucocutaneous pigmentation which can subside with age [6]. In our pediatric practice, we believe that PJS must be considered in each small bowel intussusception on a big polyp.



Figure 1: small bowel intussusception with beginner necrosis



Figure 2: Lips pigmentation

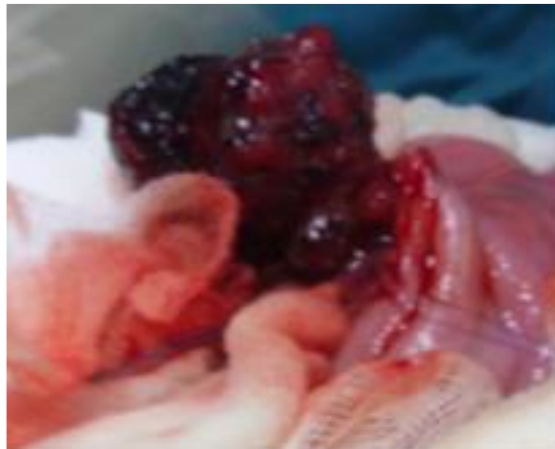


Figure 3: Intraoperative features of huge ileal polyp.

**Acknowledgment:** A written informed consent was obtained from the parents before the publication.

**Conflict of Interest:** None

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