# Peutz–Jeghers syndrome

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**ABSTRACT**

**Background:** Peutz–Jeghers syndrome (PJS) is an autosomal dominant condition presenting with mucocutaneous pigmentation. We report a rare condition of PJS in an 11-year-old female.

**Case Presentation:** An 11-year-old girl, previously healthy and developmentally normal, presented to the emergency department of the hospital with a 1-week history of vomiting and abdominal pain. She was given symptomatic treatment and referred to pediatric gastroenterology for further management and diagnosis based on the physical findings. The examination findings included hyperpigmented macules on fingertips, toes, lips, and oral mucosa. She had a history of rectal polypectomy in the past. Parents were cousins, but there was no significant family history. She was diagnosed as PJS based on her physical findings and multiple gastric polyps on endoscopy.

**Conclusion:** PJS is characterized by mucocutaneous pigmentation and multiple gastrointestinal polyps. Rectal polyp can be a clue to the syndrome even before the appearance of hyperpigmented macules. They need lifelong follow-up because they are prone to gastrointestinal complications and malignancies.

**Keywords:** Abdominal pain, gastrointestinal polyps, mucocutaneous hyperpigmentation, Peutz–Jeghers syndrome, intestinal polyposis.

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## Background

Peutz–Jeghers syndrome (PJS) is an autosomal dominant condition presenting with mucocutaneous pigmentation, gastrointestinal polyps, bleeding, intestinal obstruction, and various malignancies. The hamartomatous polyps can occur in the extraintestinal solid organs as well including lungs and renal tract. It can result in short stature and predispose the patients to various malignancies. Diagnosis is mainly clinical, but genetic analysis can be helpful in confirming the diagnosis and screening the family [1].

## Case Presentation

A 11-year-old girl, previously healthy and developmentally normal, presented to the emergency department of the hospital with a 1-week history of vomiting and abdominal pain. She was given symptomatic treatment and referred to pediatric gastroenterology for further management and diagnosis based on her physical findings (Figures 1–4). There were no other gastrointestinal symptoms. The child had a history of rectal polypectomy at the age of 3 years that presented with pain and bleeding per rectum at that time. The parents were cousins, and all siblings were normal with no significant family history.

The examination findings included hyperpigmentation of oral mucosa, lips, fingertips, and toes. Her anthropometric measurements and systemic examination were normal. She was investigated for baseline and diagnostic tests including endoscopy. Laboratories including complete blood count, urea electrolytes, stool complete examination, urine analysis, liver function test, coagulation profile, hepatic viral screening, and ultrasound abdomen were normal. Esophagogastroduodenoscopy

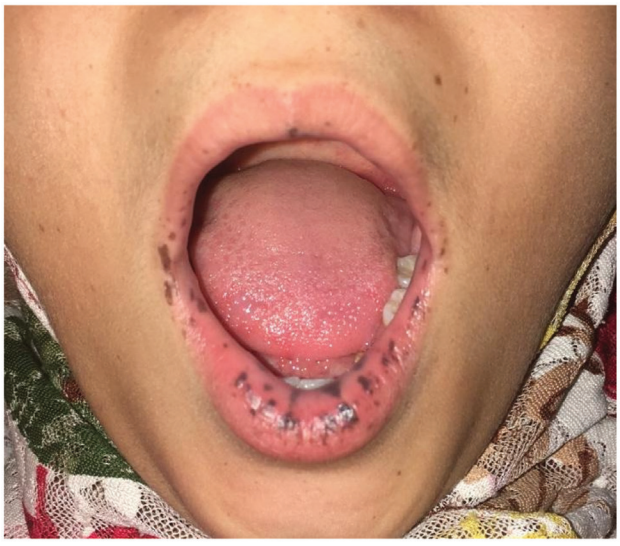

***Figure 1.*** *Hyperpigmented macules on the lips, tongue, and face.*

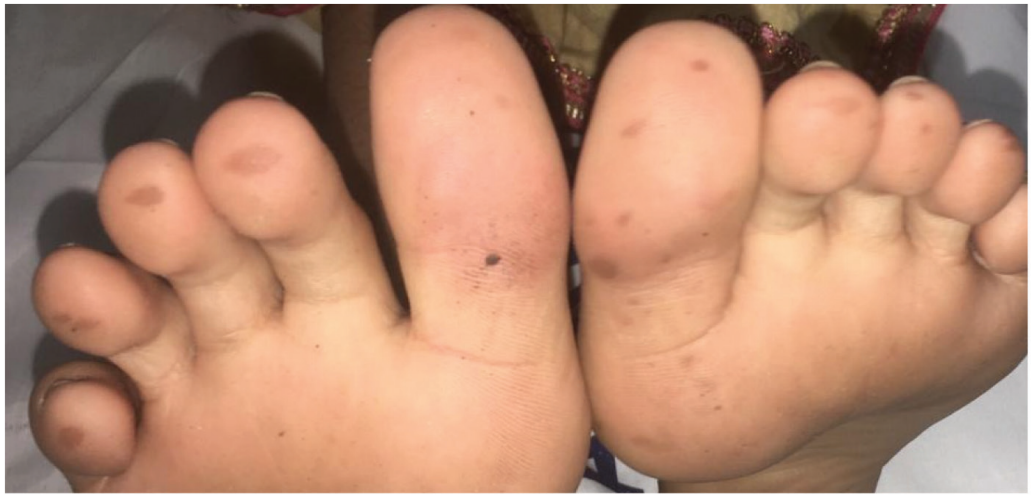

***Figure 2.*** *Hyperpigmented macules on the planter aspect of toes.*

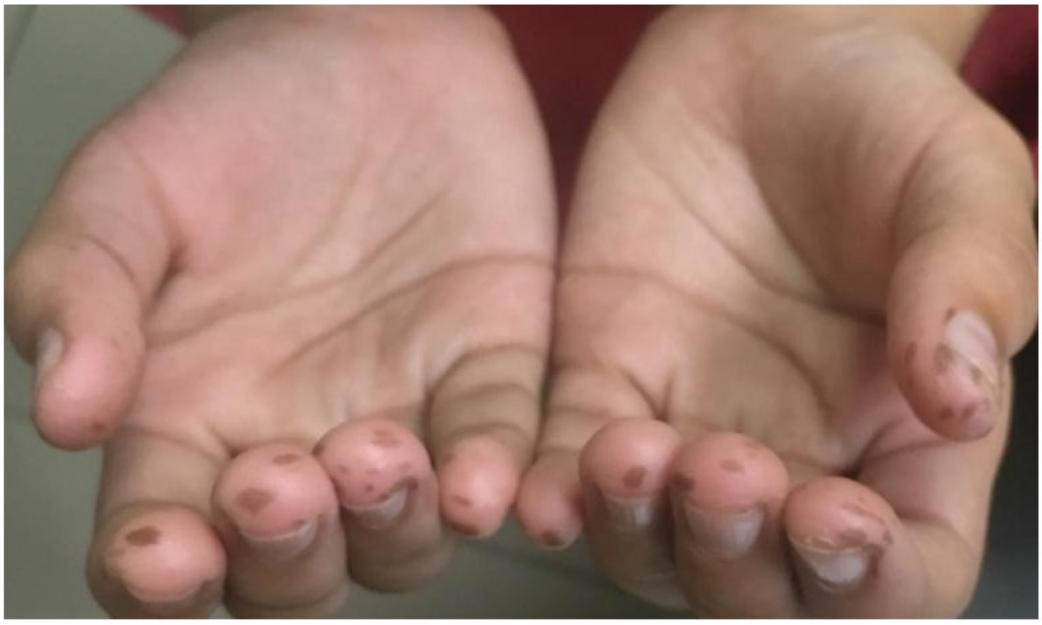

***Figure 3.*** *Hyperpigmented macules on the pulp of fingers.*

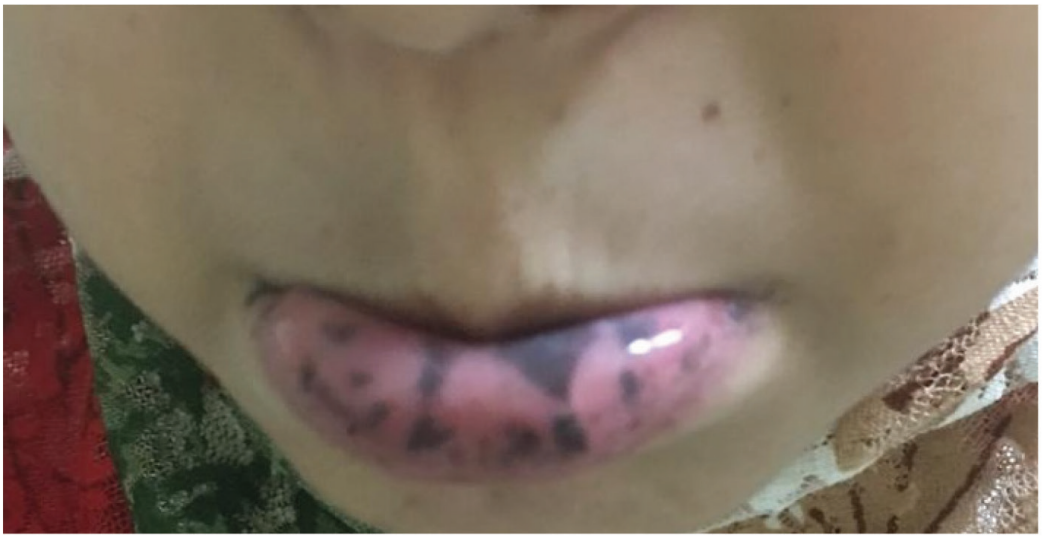

***Figure 4.*** *Hyperpigmentation on the lips.*

showed two large gastric polyps' multiple small polypoid lesions in the stomach with normal esophagus and duodenum (Figures 5 and 6). Polypectomy was planned with biopsy of the lesions. The child was called for the follow-up after 2 weeks for polypectomy and biopsy. The biopsy report showed that polypoid tissue is lined with columnar epithelium and numerous small glands. The stroma contained blood vessels infiltrated with chronic inflammatory cells comprising lymphocytes and plasma cells.

She was diagnosed as PJS based on the World Health Organization (WHO) criteria including mucocutaneous hyperpigmentation and gastric polyps [2].

## Discussion

PJS is a rare autosomal dominant disorder, and around 45% of cases have no family history and they are the result of *de novo* variant [3]. The prevalence has not been established, but according to a report, it is estimated to be 1:280,000 [4]. The clinical features include

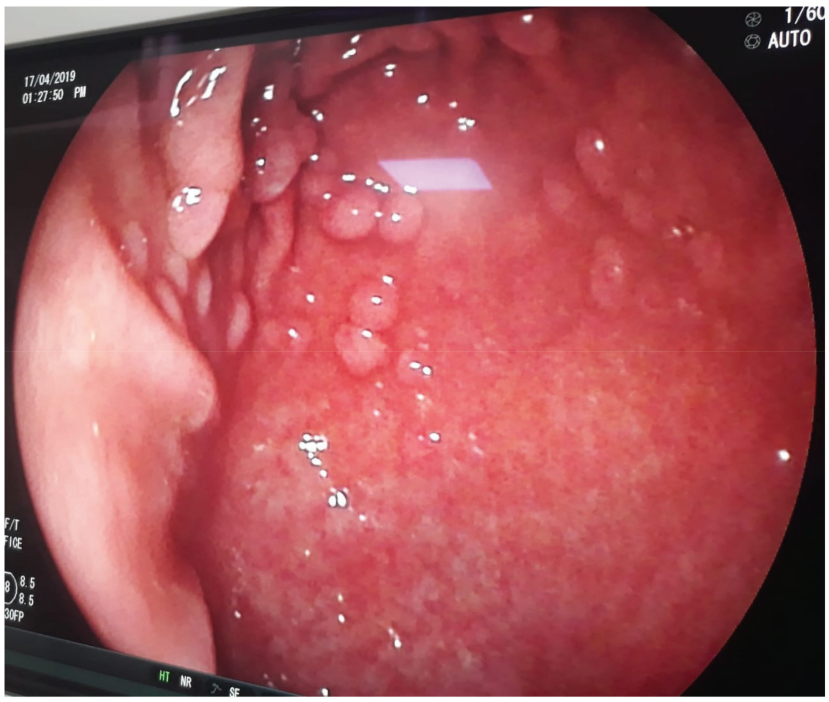


***Figure 5.*** *Multiple small fundic gland polyps in the body and fundus of the stomach.*

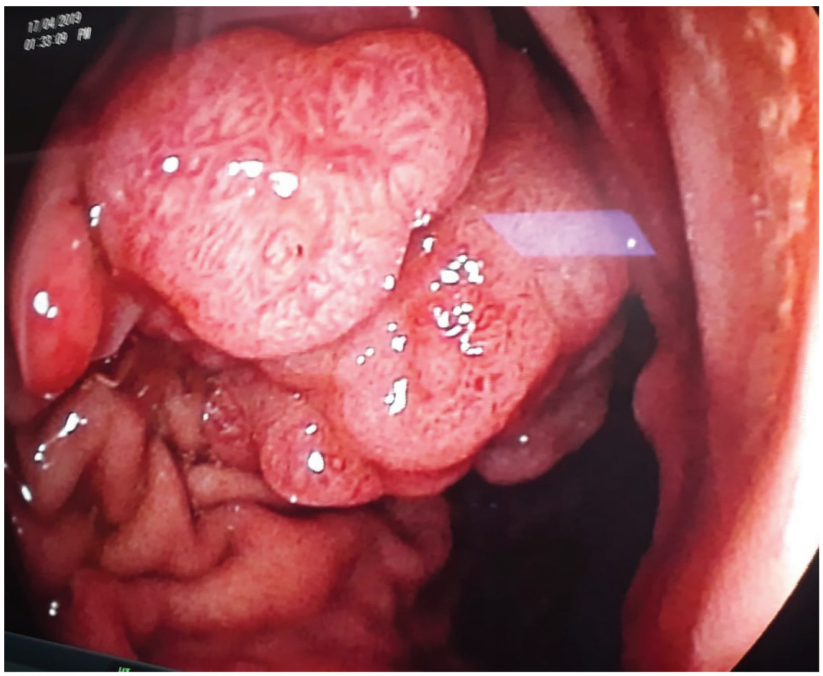

***Figure 6.*** *OGD revealed two large gastric polyps.*

mucocutaneous pigmentation and hamartomatous gastrointestinal polyps. Based on the European Consensus and WHO criteria, PJS should be suspected in any individual having intestinal polyps, mucocutaneous macules, gynecomastia in males, or history of intussusception in a child or an adult. These individuals present with abdominal pain, vomiting, gastrointestinal bleeding, and signs of obstruction ant any age. The presence of mucocutaneous macules raises the suspicion in such cases [5]. The polyps have characteristic histological findings including smooth muscle interdigitation in a characteristic tree-like pattern in the lamina propria of small bowel and lobular colonic crypts organization. The PSJ can have other kinds of intestinal polyps too which may show adenomatous changes in colonic polyps or can have hyperplastic gastric polyps [6].

The diagnosis is based on the WHO and European Consensus which says that diagnosis is established based on the following criteria [7]:

1. Three or more histologically confirmed Peutz–Jeghers polyps, or
2. Any number of Peutz–Jeghers polyps with a family history of PJS, or
3. Characteristic, prominent, mucocutaneous pigmentation with a family history of PJS, or
4. Any number of Peutz–Jeghers polyps and characteristic, prominent, mucocutaneous pigmentation

The diagnosis can be confirmed using molecular genetic testing for STK11 gene [8].

These patients are predisposed to the gastrointestinal tract, renal, genital and lung malignancies. The differential diagnosis includes juvenile polyposis syndrome which is inherited as an autosomal dominant trait and characterized by gastrointestinal polyps. Other differentials include hereditary mixed polyposis syndrome which is also inherited as autosomal dominant with colorectal polyps and predisposition to colonic tumors. Other conditions with overlapping features with PSJ are hamartoma tumor syndrome and unexplained hamartomatous mixed polyposis syndrome [9].

The patient was diagnosed as PJS based on the WHO and European Consensus criteria. The genetic analysis was not possible due to the limited resources. The child was screened for other GIT polys and malignancies. Family counseling was done on the risks and suspected future complications of disease. Family screening was planned, and the child was given follow-up for regular surveillance.

## Conclusion

Rectal bleeding due to polyps leading to polypectomy can be a clue to PJS even in the absence of skin pigmentation. It is important to screen the child for gastrointestinal tract polyps and other malignancies of lungs, genitourinary tract, and kidneys. The surveillance program, family screening for asymptomatic relatives, and genetic counseling are also of great importance in such scenarios.

### What is new?

It is a rare disease of childhood. It presents mostly as gastrointestinal symptoms such as abdominal pain, bleeding per rectum, vomiting, and hyperpigmentation. It remains a diagnostic challenge in the absence of hyperpigmentation. This case was referred for diagnosis due to the distinct physical features; otherwise, parents were not worried about the pigmentation and there was no significant family history.

### Acknowledgment

We are thankful to the patient and her family for agreeing on reporting this case.

### List of Abbreviations

| | |
|---|---|
| GIT | Gastrointestinal tract |
| ER | Emergency room |
| OGD | Esophagogastroduodenoscopy |
| PJS | Peutz–Jeghers syndrome |
| WHO | World Health Organization |

### Consent for publication

Informed consent was taken from the family.

### Ethical approval

Ethical approval is not required for publishing an anonymous case report in our institution.

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## Summary of the case

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|---|---|---|
| 1 | **Patient (gender, age)** | Female,11 years old |
| 2 | **Final Diagnosis** | Peutz–Jeghers syndrome |
| 3 | **Symptoms** | Abdominal pain and vomiting |
| 4 | **Medications** | Symptomatic |
| 5 | **Clinical Procedure** | Esophagogastroduodenoscopy, polypectomy, and biopsy of polyp |
| 6 | **Specialty** | Pediatric gastroenterology |