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Case report

Peutz–Jeghers Syndrome: A Rare Case Report with Emphasis on Oral Manifestations

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Abstract
Peutz–Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by mucocutaneous pigmentation and multiple gastrointestinal hamartomatous polyps, associated with an increased lifetime risk of malignancy. Early recognition is essential to prevent potentially serious gastrointestinal complications, including intestinal obstruction and intussusception. This case report describes a 50-year-old female who presented to the Department of Oral Medicine with multiple asymptomatic, well-defined, dark-brown to bluish-pigmented macules involving the lower lip, buccal mucosa, and palate. The patient had a previous history of gastrointestinal symptoms and intestinal polyps. Gastrointestinal endoscopic examination revealed multiple polyps, while histopathological assessment demonstrated characteristic hamartomatous polyps with arborizing bundles of smooth muscle. A family history of gastrointestinal polyposis was also reported, supporting the hereditary nature of the condition. The clinical findings were consistent with Peutz–Jeghers syndrome, which is associated with pathogenic variants in the STK11 tumor suppressor gene. This case highlights the importance of recognizing characteristic oral and perioral pigmentation as a potential indicator of an underlying systemic disorder. Dental and oral medicine practitioners play an important role in the early identification and referral of suspected cases for appropriate gastrointestinal evaluation, genetic counseling, and lifelong surveillance to reduce the risk of associated complications and malignancies.
Keywords: Peutz–Jeghers Syndrome; Mucocutaneous Pigmentation; Oral Pigmentation; Hamartomatous Polyp.

Introduction

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant inherited disorder that always requires early diagnosis. It is characterized by the presence of hamartomatous polyps in the gastrointestinal tract, typically found in the small intestine and colon.[1] PJS patients have an increased risk of malignancy compared with the general population [2]. Thus, they require long-term follow-up to assess the diagnosis, treatment of malignant changes, and other severe complications secondary to Peutz-Jeghers syndrome.

Considering the recent advances in treatment, early management of PJS individuals is essential to avoid an unnecessary surgical emergency. Recurrent intestinal obstruction, particularly due to small bowel intussusception in younger patients, represents a serious complication [3].

We report a new familial case and highlight the clinical, histopathological, and therapeutic features of this syndrome, whose pathogenesis remains incompletely understood.

Case Presentation

A 50-year-old female patient presented to the Department of Oral Medicine, Faculty of Dentistry, Zawia University, with multiple asymptomatic pigmented macules involving the lips and oral mucosa.

Clinical examination revealed multiple well-defined, flat, dark-brown to bluish macules predominantly involving the lower lip, buccal mucosa, and palate. The patient reported a history of gastrointestinal symptoms and previously diagnosed intestinal polyps.

Based on the characteristic mucocutaneous pigmentation and gastrointestinal findings, Peutz–Jeghers syndrome was suspected. Gastrointestinal endoscopic examination demonstrated multiple polyps, and histopathological examination revealed hamartomatous polyps with characteristic arborizing bundles of smooth muscle. Genetic testing, when available, demonstrated a pathogenic variant in the STK11 gene.

Discussion

Peutz–Jeghers syndrome is inherited in an autosomal dominant fashion, with non-sporadic cases typically demonstrating a history in a first-degree relative, as in our patients whose father was affected. It is characterized by

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the presence of pigmented macules on the lips, oral mucosa, and digits, along with multiple hamartomatous polyps throughout the gastrointestinal tract [4]. The condition is caused by pathogenic variants in the STK11/LKB1 tumor suppressor gene located on chromosome 19p13.3, which plays a crucial role in cell growth regulation and tumor suppression. Familial aggregation is common, as illustrated in our cases, where the father had a history of colonic polyposis. [5,6]



Figure 1. Multiple pigmented macules on the lips, buccal mucosa and palate associated with Peutz-Jeghers syndrome

The diagnosis of PJS is characteristic of childhood or early adulthood. The male-to-female ratio for PJS is almost 1:1. Pigmentation is typically observed in childhood, while it may even disappear in adulthood.[6] Our patient has not presented any pigmentation since she remains under our observation. Therefore, this case falls under the first category of WHO criteria for PJS diagnosis. Polyps in the outcome of Peutz-Jeghers syndrome can occur anywhere in the gastrointestinal tract [7].

The present case is of particular interest because the patient presented to an oral medicine clinic, highlighting the important role of oral healthcare professionals in the early recognition of systemic diseases. Characteristic mucocutaneous pigmentation is a well-recognized feature of PJS and may be among the earliest clinical manifestations of the syndrome. These lesions typically appear as multiple, small, well-demarcated, brown to blue-black macules involving the lips, perioral skin, and oral mucosa, particularly the buccal mucosa, gingiva, and palate. Pigmentation may develop during childhood and can precede the diagnosis of gastrointestinal polyposis by several years [8]. Consequently, the identification of characteristic oral pigmentation should prompt a comprehensive medical history and further systemic investigation, even in patients without apparent gastrointestinal symptoms.

Conclusion

Peutz–Jeghers syndrome is an uncommon but clinically important condition in which oral manifestations may precede gastrointestinal complications. Recognition of characteristic oral pigmentation by dental professionals can facilitate early referral, diagnosis, genetic counseling, and appropriate lifelong surveillance.

Declarations

Ethical Approval

Ethical approval was obtained from the relevant institutional ethics committee, where applicable. The study was conducted in accordance with the principles of the Declaration of Helsinki.

Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and the accompanying clinical photographs.

Availability of Data and Materials

All data supporting the findings of this case report are included in the manuscript.

Competing Interests

The authors declare that they have no competing interests.

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