ISSN 1596-3519

Volume 20 Issue 4 October-December 2021

www.annalsafrmed.org

# Annals of African Medicine

Wolters Kluwer

Medknow

Case Report

# Solitary Peutz–Jeghers Type Hamartoma in a Nigerian: A Case Report of a Rare Finding and Review of Literature

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## Abstract

**Background:** We report a case of solitary Peutz–Jeghers (P-J) type hamartomatous polyp in the sigmoid colon of an asymptomatic Nigerian without other diagnostic criteria for full-blown P-J syndrome. **Case Report:** During colonoscopy for a 58-year-old man, a solitary, pedunculated polyp was seen in the sigmoid colon. Histopathological examination of the endoscopically resected mass revealed the classical features of P-J type hamartoma. A search for lentigines on the skin or mucous buccal membranes was negative. There is no family history of such findings. **Conclusion:** The case here presented is truly rare as a review of the scientific literature appears, to the best of our knowledge, not to contain such a unique presentation from our locality nor indeed from among Black Africans. We conducted a review of the literature and wished to highlight the evolving concept that solitary P-J polyps be considered a distinct disease entity when it appears in the absence of other features of the syndrome complex.

**Keywords:** Colonoscopy, mucocutaneous pigmentation, Nigeria, Peutz–Jeghers syndrome, solitary Peutz–Jeghers hamartoma

## Résumé

**Contexte:** Nous rapportons un cas de polype hamartomateux solitaire de type Peutz – Jeghers (P J) dans le côlon sigmoïde d'un Nigérian asymptomatique sans autres critères diagnostiques du syndrome P J complet. **Rapport de cas:** Au cours de la coloscopie d'un homme de 58 ans, un polype solitaire pédonculé a été observé dans le côlon sigmoïde. L'examen histopathologique de la masse réséquée par voie endoscopique a révélé les caractéristiques classiques de l'hamartome de type PJ. Une recherche de lentigines sur la peau ou les muqueuses buccales a été négative. Il n'y a pas d'antécédents familiaux de telles découvertes. **Conclusion:** Le cas présenté ici est vraiment rare car une revue de la littérature scientifique semble, à notre connaissance, ne pas contenir une présentation aussi unique de notre localité ni même des Noirs africains. Nous avons effectué une revue de la littérature et avons souhaité mettre en évidence le concept évolutif selon lequel les polypes P J solitaires doivent être considérés comme une entité pathologique distincte lorsqu'ils apparaissent en l'absence d'autres caractéristiques du complexe du syndrome.

**Mots clés:** Coloscopie, pigmentation mucocutanée, Nigéria, syndrome de Peutz – Jeghers, hamartome de Peutz – Jeghers solitaire

## Introduction

Peutz–Jeghers syndrome (PJS) is an autosomal dominant, hereditary disease characterized by hamartomatous polyps of the gastrointestinal (GI) tract and by mucocutaneous melanin deposits.[1] Its cause, in most cases (>90%), appears to be a germline mutation of the serine/threonine kinase 11 (STK11/LKB1) tumor suppressor gene, located on chromosome 19p13.[2-4] It is associated with an increased risk

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Quick Response Code:

Website: www.annalsafrmed.org

DOI: 10.4103/aam.aam_37_20

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**How to cite this article:** Oluyemi AO, Odeghe EA, Awolola NA. Solitary peutz–jeghers type hamartoma in a Nigerian: A case report of a rare finding and review of literature. Ann Afr Med 2021;20:307-9.

**Submitted:** 07-May-2020 **Revised:** 27-Jun-2020
**Accepted:** 07-Aug-2020 **Published:** 03-Dec-2021

of developing cancers, both GI and non-GI, with many patients developing cancer at a young age.[5-7]

The World Health Organization diagnostic criteria for PJS include any of the following:[8]

- Three or more histologically confirmed Peutz–Jeghers (P-J) polyps, or
- Any number of P-J polyps with a family history of P-JS, or
- Characteristic, prominent, mucocutaneous pigmentation with a family history of PJS, or
- Any number of P-J polyps and characteristic, prominent, mucocutaneous pigmentation.

Recently, there have been a number of case reports of solitary GI (gastric, small intestinal, and colorectal) hamartomatous polyps that are histologically similar to those of the P-JS type but without the typical mucocutaneous lesions or a family history of PJS.[9,10] These are called solitary P-J polyps. Some of these have been associated with complications that are seen with PJS such as intussusception and recurrent small bowel and biliary obstruction.[10-12]

The scientific literature does contain some reports of PJS in Sub-Saharan Black Africans, but these are sparse indeed – thus giving credence to the as-yet unproven supposition that the disease entity is a rarity over here.[13-15] The documented cases of solitary P-J type polyps without evidence of mucocutaneous involvement are rare. This case report documents one such case. We also looked at some other aspects of the genetic basis and diagnosis of this evolving clinical entity.

## Clinical Case

A 58-year-old asymptomatic man presented for routine screening colonoscopy. A solitary, pedunculated polyp (Paris Classification Ip) was seen in the sigmoid colon [Figure 1]. The lesion was excised through polypectomy, and the sample was sent for histopathologic diagnosis. A subsequent check for any pigmentation in the mucocutaneous membranes of the man and those of his two siblings that were on hand was negative.

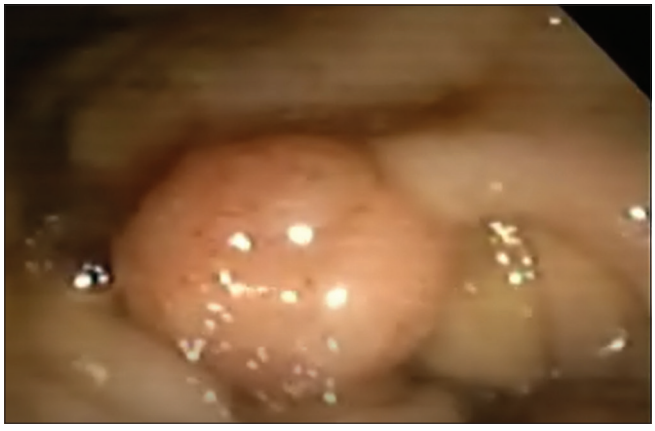

**Figure 1:** The endoscopic view of the polyp in the sigmoid colon

The histopathology results described the typical findings of P-J hamartomatous polyps by the extensive smooth muscle proliferation, with an elongated, arborized pattern throughout the lesion [Figure 2]. A repeat of the colonoscopy was carried out 2 years later, but this was normal, as was that of the said siblings.

## Discussion

The endoscopic appearance of these lesions is in no way specific to this condition; thus, the histopathology of the polyps is of paramount diagnostic importance.[16] The pathological feature of solitary P-J type polyp is that the smooth muscle bundles from the muscularis mucosae extend from a core to the entire polyp and form a typical branch-like structure covered by almost normal mucosa.[8,9] The diagnosis of P-J polyps is usually based on the finding of this characteristic pattern. However, Tse *et al*. argue that the presence of a lobulated growth pattern be added to the presence of the earlier arborizing smooth muscle pattern before the diagnosis is reached.[17]

A case series done in eight patients with solitary P-J polyps found that these patients were older at the time of diagnosis than those with PJS, and on follow-up for a median of 11.5 years, none of them developed another P-J polyp or cancer.[18] This led the authors to conclude that patients with solitary P-J polyps do not have an increased risk of cancer development. However, other studies have reported the presence of both GI and extra-GI cancers in some patients with solitary P-J polyps, including adenomatous transformation and malignant transformation of some of the polyps.[19-21]

Genetic analysis of mutations of the STK11/LKB1 gene in many of these patients with solitary P-J polyp did not show any genomic abnormality.[22,23] However, this is to be paralleled

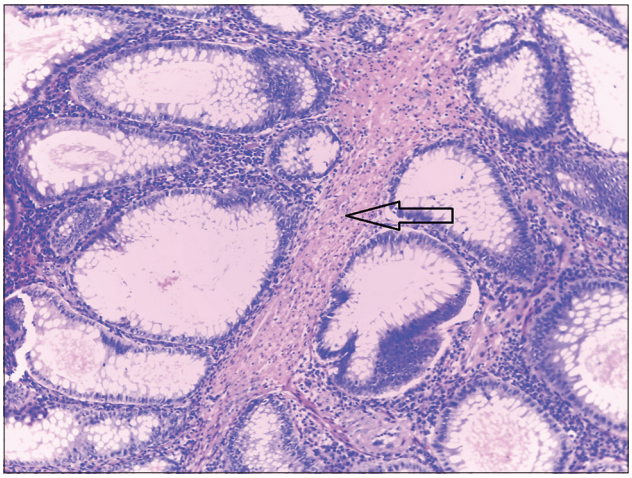

**Figure 2:** Histopathologic picture of the colonic PeutzJeghers type hamartoma polyp. A medium-power photomicrograph that clearly depicts the characteristic central core of branching smooth muscle (transparent arrow), H and E, ×100

with the fact that such mutations are not demonstrable in some 10% of polyps from PJS-affected individuals as well.[24]

The differentiating findings of a lack of mucocutaneous features, absence of a family history, absence of an increased risk of developing cancers, and absence of the genetic mutation, have led some authors to question if they may be different disease entities.

Even though many of these patients with solitary P-J polyps do not have genetic mutations in the STK11 gene, a recent study found the presence of epigenetic abnormalities in the STK11/LB gene.[25] This may mean that PJS and solitary P-J polyps are the results of different mutation pathways, and therefore may be distinct diseases with similar histopathological expression.

## Conclusion

This case of solitary P-J type hamartoma in the absence of other components to describe it as PJS is unique in our locality as it is, to the best of our knowledge, the first from Nigeria. It also presents a short review of various documented genetic, clinical, and pathological aspects of this newly emerging distinct disease. The article highlights the current thinking that solitary colonic P-J polyps may be a distinct entity from PJS.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

### Financial support and sponsorship

Nil.

### Conflicts of interest

There are no conflicts of interest.

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