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Phacomatosis Pigmentokeratotica with Atypical Features in A 10-Year-Old Yemeni Boy: A Case Report

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Abstract

We report a case involving a 10-year-old Yemeni boy who presented with multiple linear sebaceous nevi on the left side of the scalp, face, and anterior neck, accompanied by a left-sided papular nevus spilus affecting the same regions. Moreover, the patient exhibited atypical and previously unreported features, including melanocytic lesions on the left oral mucosa and superimposed melanocytic lesions over the nevus sebaceous in the parieto-occipital area. Notably, most facial melanocytic lesions were hairy, with no evidence of extracutaneous involvement.

1. Introduction

Phacomatoses, i.e., neurocutaneous disorders, represent a heterogeneous group of congenital conditions characterized by various nevus and hamartoma types [1]. To define this group, the term 'phacomatosis' (derived from the Greek word phakos, meaning 'birthmark') was first proposed by the Dutch ophthalmologist van der Hoeve in 1923[2]. Currently, scientific and medical communities distinguish two phacomatosis types, i.e., namely phacomatosis pigmentovascularis and pigmentokeratotica (PPV and PPK, respectively), [3] the latter having been defined by Happle et al. in 1996 as a distinct Epidermal Nevus Syndrome (ENS) type, characterized by Sebaceous Nevus (SN) and Papular Speckled Lentiginous Nevus (PSLN) co-occurrence [3]. As per the twin spot hypothesis, two homozygous recessive mutations could explain epidermal and melanocytic nevus coexistence, denoting a putative PPK paradigm in humans [4].

2. Case report

A 10-year-old Yemeni boy presented with multiple alopecic areas as well as black melanocytic nevi on the scalp as well as the face and neck, respectively, having persisted since birth. Cutaneous examination revealed two linear, yellow, greasy alopecic plaques on the parieto-temporal and parieto-occipital areas of the left side, one extending into the preauricular area, superimposed by broad black patches (Figure 1a–d). The left side of the patient's face was covered with multiple black lesions in the diameter range of 2–20 mm, varying from flat to elevated, on a light brown background. Coarse black hair covered most lesions, a linear, yellow, plaque being present on the median line of the anterior neck. Furthermore, maculopapular black lesions covered the left side of both lips and the oral mucosa.

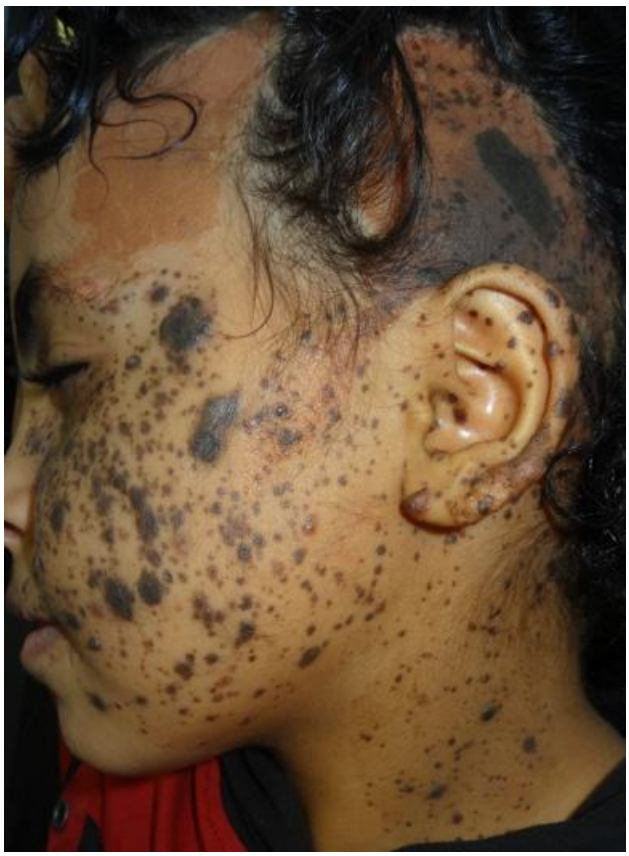

Figure 1a: Left lateral view. Papular nevus spilus and sebaceous nevus. Note the overlapping melanocytic nevus within the sebaceous nevus in the parieto-occipital area that extends to the preauricular area. The parieto-temporal NS is not involved and extends to the lateral part of the left eyebrow.

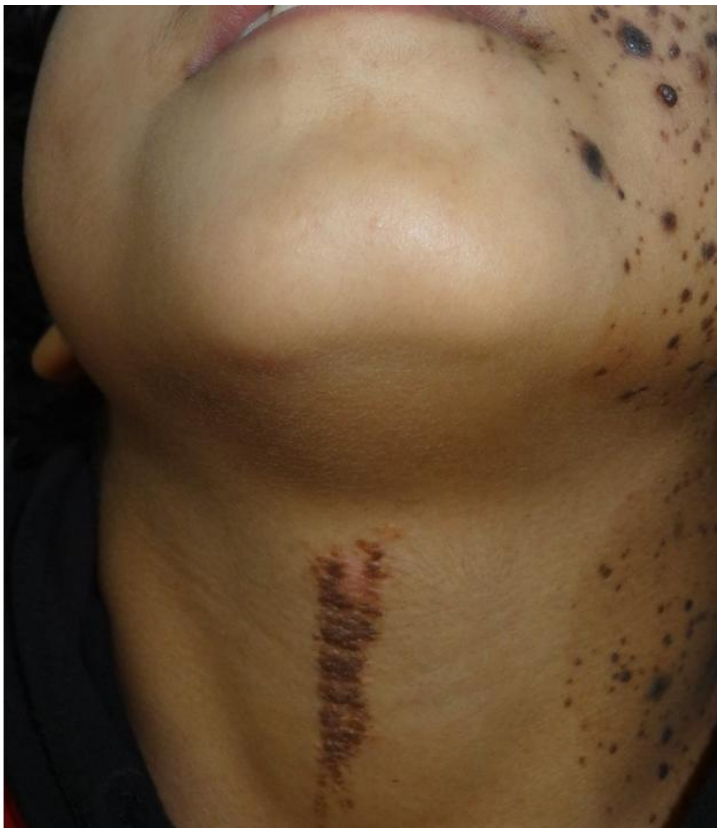

Figure 1b: Anterior view of the median linear NS on the anterior neck and a light brown background of the papular nevus spilus.

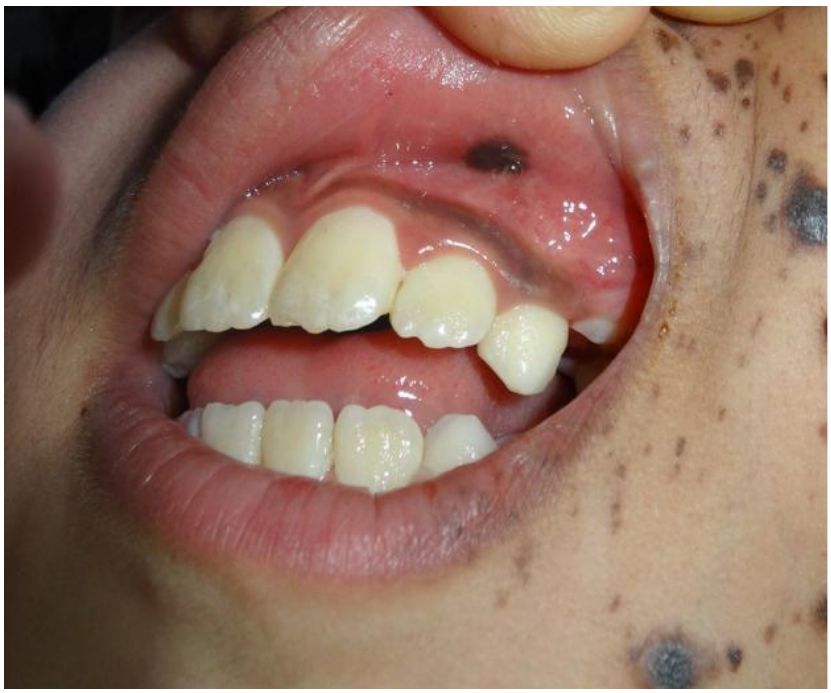

Figure 1c: Involvement of the left side of the oral mucosa with papular melanocytic nevi.

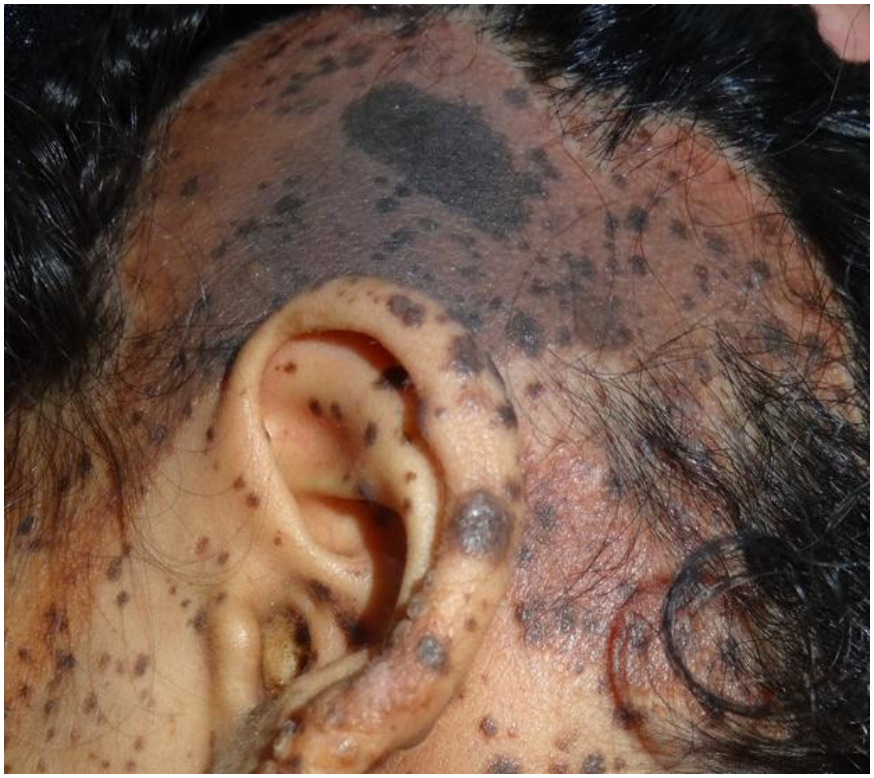

Figure 1d: A close-up view of the parieto-occipital NS, extending to the pre-and postauricular areas, intermingled with papular melanocytic lesions.

At presentation, the presence of large sebaceous nevi on the scalp raised suspicion for sebaceous nevus syndrome, or Schimmelpenning-Feuerstein-Mims- syndrome (SFMS), a subtype of Epidermal nevus syndrome. However, the diagnosis was considered less likely due to the absence of other characteristic features of SFMS, such as coloboma, lipodermoid of the conjunctiva, and the presence of a papular speckled nevus on the left side of the face.

The patient also exhibited atypical, previously unreported features, including melanocytic lesions on the left oral mucosa and melanocytic lesions superimposed on the nevus sebaceous in the parieto-occipital region. Notably, most facial melanocytic lesions

were associated with coarse hair growth. There was no family history of a similar condition (Table 1). Punch biopsies of the scalp plaques revealed histopathological features consistent with SN. Genetic testing for HRAS or KRAS mutations could have confirmed the diagnosis but was unavailable due to geographical limitations. Based on characteristic clinical and dermatopathological findings, a diagnosis of PPK was established. Surgical excision of the SN was planned due to the potential risk of malignant transformation. To our knowledge, this is the first reported case of PPK in Yemen.

Table 1: Summary of clinical features in the present case compared with previously reported PPK case

Clinical Feature	PPK	Present case
Sebaceous nevus	+	+
Papular nevus spilus	+	+
Mucosal melanocytic nevi	_	+
Melanocytic nevi on sebaceous nevus	_	+
Coarse hair over melanocytic lesions	_	+

6. Discussion

ENS is an umbrella term encompassing syndromes characterized by epidermal nevi accompanied by both cutaneous and extracutaneous abnormalities. Based on the variant of epidermal nevus, ENS can be classified into organoid syndromes, including PPK, Schimmelpenning syndrome, nevus comedonicus, angora hair nevus, and Becker nevus syndromes, and keratinocytic nevus syndromes [5]. PPK is caused by postzygotic mutations in the HRAS or KRAS genes and is thus considered a mosaic RASopathy, characterized by both cutaneous and extracutaneous manifestations [6]. If these mutations occur during embryogenesis, they may lead to a variety of extracutaneous manifestations. Most patients with PPK present with extracutaneous features, most commonly involving the ocular (strabismus and ptosis), neurologic (hemiparesis, hyperhidrosis, and dysesthesia), and musculoskeletal (scoliosis and muscular weakness) systems [7]. Similar to the present case, fewer than 30 % of reported PPK cases (n = 30) lack extracutaneous involvement.

This report describes a case of PPK with atypical features, contributing to the limited literature on this extremely rare genodermatosis. Notably, this case highlights a rare presentation of PPK without extracutaneous involvement. Patients with cutaneous PPK are at increased risk of malignant transformation of sebaceous nevi into basal cell carcinoma and of papular-speckled lentiginous nevi into melanoma. Therefore, regular clinical monitoring is essential for such patients [8]. Although PPK has been associated with nondermatological malignancies such as rhabdomyosarcoma and nephroblastoma (Wilms tumor), as observed in other RASopathies, no internal malignancies have been reported in PPK patients without extracutaneous involvement [8]. Although there is currently no effective medical therapy for PPK, the patient in this case exhibited a favorable response to long-pulsed alexandrite laser treatment for the pigmented facial lesions. In contrast, the SN lesions were referred for surgical excision. The psychosocial impact of visibly extensive and cosmetically distressing lesions should always be considered during treatment planning, as such conditions can significantly affect patients' quality of life.

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