Congenital Triangular Alopecia Associated with Phakomatosis Pigmentovascularis Type II along with Klippel Trenaunay Syndrome

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Abstract

Phakomatosis pigmentovascularis (PPV) is characterized by the association of a vascular nevus with a pigmentary nevus and is divided into five subtypes. PPV type II or Happle's phakomatosis cesioflammea is the most common subtype comprising of nevus flammeus along with pigmentary nevus in the form of aberrant Mongolian spots, nevus of Ota or less frequently nevus of Ito. It is estimated that around 50% of patients with PPV have systemic involvement, most frequently involving the central nervous system and eye. Other associated features include vascular abnormalities such as Sturge-Weber syndrome, and klippel trenaunay syndrome (KTS), and cutaneous lesions such as nevus anemicus (most common), cafe'-au-lait macules, generalized vitiligo and congenital triangular alopecia (CTA). There are only four reports of PPV associated with CTA in literature, and only a single previous report with associated KTS and this association has not been reported previously from India. We describe a case of a 30-year-old male having phakomatosis pigmentovascularis type II along with klippel trenaunay syndrome and associated with congenital triangular alopecia.

Keywords: *Congenital triangular alopecia, klippel trenaunay syndrome, mosaicism, phakomatosis pigmentovascularis*

Introduction

Phakomatoses pigmentovascularis (PPVs) are rare syndromes characterized by the coexistence of a vascular malformation with a pigmentary nevus. PPV is divided into five subtypes based on the type of pigmented and vascular lesion. PPV type II or Happle's phakomatosis cesioflammea is the most common subtype comprising of nevus flammeus along with dermal melanocytoses such as aberrant Mongolian spots, nevus of Ota or uncommonly nevus of Ito. Phakomatosis cesioflammea may be associated with other cutaneous lesions with nevus anemicus being the most common, present in 50% of the patients in one series. Other cutaneous lesions which have been reported with PPVs include cafe'-au-lait macules, generalized vitiligo and rarely congenital triangular alopecia (CTA). We describe a case of PPV type II associated with CTA which has not been reported previously from India.

Case Report

A 30-year-old male presented to us with bluish-gray discoloration around the right eye and the trunk and red macules and plaques on the trunk and extremities, since birth. A triangular patch of hairloss was also present on the right side of the scalp above the ear since birth [Figure 1a and b]. The patient was otherwise healthy and family history was unremarkable. Cutaneous examination revealed multiple erythematous patches suggestive of capillary malformations involving the neck, trunk and extremities with hemihypertrophy of the right hand and foot. Multiple bluish-grey macules suggestive of aberrant Mongolian spots were present on the trunk [Figure 2a and b]. Bluish-brown macules suggestive of nevus of Ota were present involving the right periorbital region and sclera. Also, dilated veins were present on the antero-medial aspect of right leg. A well circumscribed triangular area of non-scarring non-inflammatory hairloss with few vellus hair was noted on the right temporal area. Trichoscopy of the alopecic patch revealed few vellus hair of varying lengths [Figure 3a and b]. Skin biopsy form this area revealed decreased number of follicular units with a single vellus hair [Figure 4a and b]. There were

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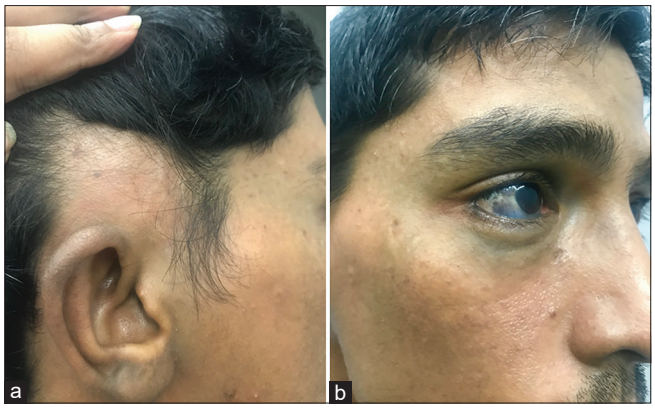

Figure 1: (a) A well circumscribed triangular area of non-scarring hair loss with a few vellus hair involving the right temporal area. (b) Nevus of Ota in the right periorbital region along with involvement of sclera of right eye

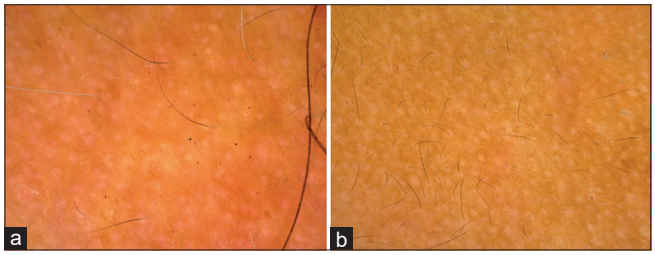

Figure 3: (a and b) Trichoscopy of the alopecic patch revealed few vellus hair of varying lengths (50x, polarized mode, Dinolite AM 4113 ZT)

no associated cardiac, central nervous system (CNS), dental and muculoskeletal abnormalities. Venous doppler ultrasonography of the limbs revealed a dilated great saphenous vein from the thigh to the ankle with normal deep venous and lymphatic system. Based on the above clinical, histological and dermoscopic findings, a diagnosis of CTA associated with PPV type II and klippel trenaunay syndrome (KTS) was reached. The prognosis of the condition was explained to the patient, and the treatment options including Nd-YAG laser for nevus of ota and pulse-dye laser for capillary malformations were offered. However, he refused therapy and was lost to follow-up.

Discussion

PPVs are believed to arise due to aberrant development of blood vessels, and abnormal melanocytic migration during embryogenesis.[1] It is postulated that loss of heterozygosity leads to nonallelic twin spotting, that is, presence of two distinct mutant tissues, different from the surrounding normal tissue, resulting in two adjacent cutaneous lesions. Around 50% of PPV patients have systemic involvement, most frequently involving the CNS and eye. Moreover, vascular abnormalities such as Sturge-Weber syndrome and KTS and cutaneous lesions such as nevus anemicus, cafe'-au-lait macules, generalized vitiligo and CTA may co-exist as well.

CTA also known as temporal triangular alopecia, presents as an asymptomatic non-cicatricial circumscribed patch of alopecia usually located in the front otemporal region

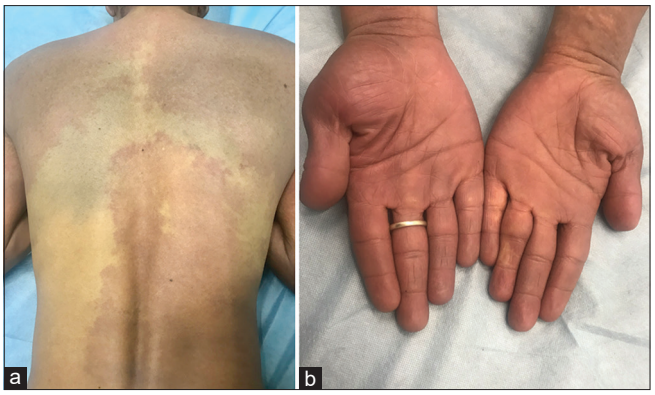

Figure 2: (a) Capillary malformations and multiple bluish grey macules suggestive of aberrant Mongolian spots involving the neck, trunk and extremities. (b) Hemihypertrophy of the right hand

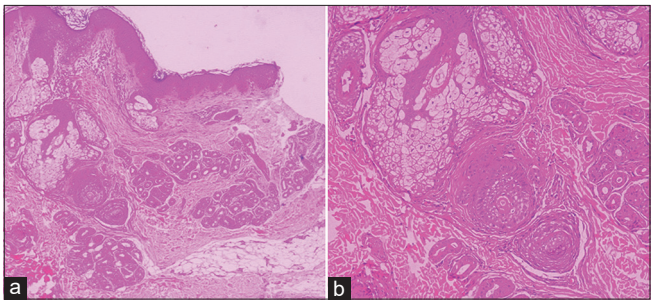

Figure 4: (a) Scalp biopsy showing marked reduction of hair follicles with single vellus follicular unit. (H and E, ×40) (b) Vellus follicle at higher magnification. (H and E, ×100)

with <20% present bilaterally.[2] The lesions are usually present at birth or develop during the first few years of life and remain stable thereafter. Majority of CTA cases are completely bald, though a few normal vellus or terminal hair may be present within the affected patch. The lesions of CTA are characterized histologically by miniaturization of the hair follicles and replacement of the terminal hair with sparse vellus hair and dermoscopically by the presence of few vellus hair of varying length with absence of specific features suggestive of other causes of localized alopecia.

In 15% cases, CTA may be associated with systemic disorders such as congenital heart diseases, CNS, bone and teeth abnormalities, and rarely it can be a part of malformation syndromes such as PPV. Our patient presented with lesions classical of PPV type II and KTS along with CTA on the right side. There are only four reports of PPV with CTA in literature, and only a single previous report with associated KTS.[3-6] Further, the co-existence has not been reported previously from India.

The exact etiology of CTA remains unclear; some believe that CTA occurs because of mosaicism with a paradominant inheritance pattern and postzygotic loss of wild type allele in a heterozygote state manifests the disease. Others suggested that CTA may be a localized ectodermal defect or an epidermal nevus. Our case, along with previous reports of CTA present in association

with rare syndromes such as PPV, suggest it may be an additional feature rather than a coincidental finding. Though PPV usually manifests with pigmentary and vascular mosaicism, it seems plausible that there may be a broader underlying ectodermal mosaic defect at times, explaining the occasional involvement of other ectoderm derivatives like hair follicles, eye and CNS. Further research on the pathogenesis of the condition including genetic studies to identify the causative genes should be considered to clarify this aspect further.

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.

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Conflicts of interest

There are no conflicts of interest.

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