# Phacomatosis cesioflammea with Klippel Trenaunay syndrome: A rare association

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## ABSTRACT

A 30-year-old Indian male presented with bilateral Nevus of Ota, extensive nevus flammeus over the trunk and left lower limb with soft tissue hypertrophy and varicosities affecting the left lower limb. He was otherwise in good general health. A diagnosis of Phacomatosis cesioflammea or Phacomatosis pigmentovasularis Type II with Klippel Trenaunay syndrome was made. The case is being reported on account of its rarity.

**Key words:** Klippel Trenaunay syndrome, Nevus of Ota, Phacomatosis cesioflammea, Phacomatosis pigmentovascularis.

## INTRODUCTION

Phacomatosis cesioflammea, earlier called as Phacomatosis pigmentovasularis (PPV) Type II is a rare condition characterized by the coexistence of dermal melanocytosis (Mongolion spot or Nevus of Ota) and nevus flammeus.[1] Klippel Trenaunay syndrome (KTS) manifests as asymmetric overgrowth of a limb and soft tissue associated with vascular malformation (nevus flammeus) and varicosities.[2] We report a case that illustrates a rare association between phacomatosis cesioflammea and KTS.

## CASE REPORT

A 30-year-old male presented with extensive vascular changes on the trunk and limbs and pigmentary changes on the face. Cutaneous examination showed extensive nevus flammeus affecting almost the entire back, crossing the midline and extending to the buttocks and left lower limb [Figure 1a and b]. Similar discrete lesions were present over the flanks and lateral aspect of the chest [Figure 2]. There was bilateral bluish-grey pigmentation over the face along the ophthalmic and maxillary division of the trigeminal nerve consistent with Nevus of Ota [Figure 3a]. The sclera, nasal mucosa and palate also showed patchy bluish-grey pigmentation bilaterally [Figure 3a and b]. Soft tissue hypertrophy of the left lower limb was seen in association with nevus flammeus and varicose veins over the left leg. A difference of 3 cm was noted in the circumference of both lower legs at the mid-calf region. The patient also had right-sided scoliosis. Neurological and ophthalmic examinations did not reveal any abnormality. Routine hematological investigations were normal. Radiological examination of the left lower limb showed soft tissue swelling without evidence of bony changes. Color Doppler of the affected limb showed varicosities of short saphenous vein with incompetence of the perforators. There was no history of seizures. Also, there was no history of similar pigmentary or vascular lesions in any of his family members.

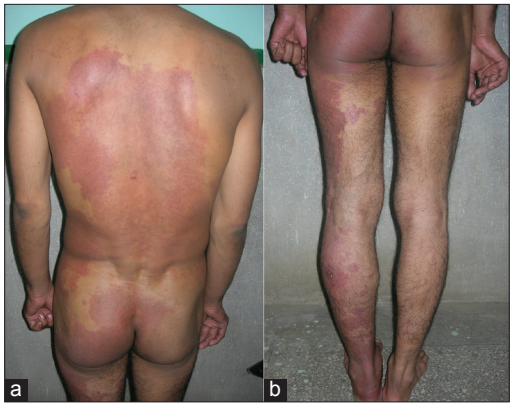


**Figure 1:** (a) Showing extensive nevus flammeus over the back, crossing the midline and extending to the buttocks and left lower limb (b) Showing hypertrophy of left lower limb with nevus flammeus and varicose veins

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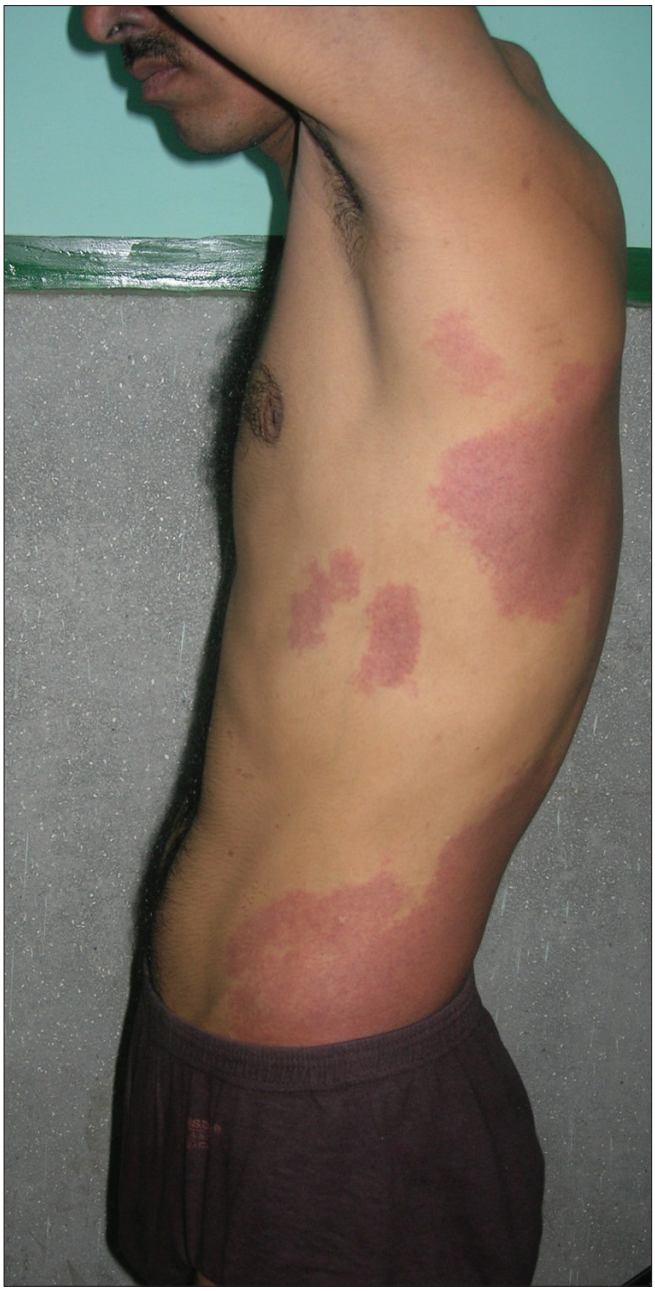

**Figure 2:** Showing nevus flammeus over the flanks and lateral aspect of the chest (left side)

## DISCUSSION

PPV is a rare disorder that exhibits an association of a widespread vascular nevus with pigmentary nevus.[3] It was first described by Ota in 1947.[4] The term "Phacomatosis" was originally used to characterize some neuro-cutaneous syndromes, but is now mainly applied to genetically determined diseases characterized by the presence of two or more different nevi such as PPV.[2]

Hasegava[3] classifies PPV into four types. Each type involves a nevus flammeus with an additional nevus, and is further divided into subtypes a and b. Subtype "a" has cutaneous involvement only while subtype "b" has extracutaneous involvement. Cutis marmorata telangiectasia congenital (CMTC) with Mongolian spot have been reported as type V, a distinct variant of PPV.[5] In 2005, Happle[1] proposed a newer and simplified classification scheme for PPV including three different categories [Table 1]. Phacomatosis cesioflammea (blue spots with nevus flammeus, traditional type IIa/IIb), phacomatosis spilorosea (nevus spilus with pale pink telangiectatic nevus, traditional type IIIa/IIIb) and phacomatosis cesiomarmorata (blue spots with cutis marmorata telangiectasia congenita, traditional type V). He also added a category of unclassifiable forms that cannot be included into the rest of the three groups. The traditional type I was dropped in this classification as it was extremely rare and virtually non-existent. Phacomatosis cesioflammea is the most frequently occurring

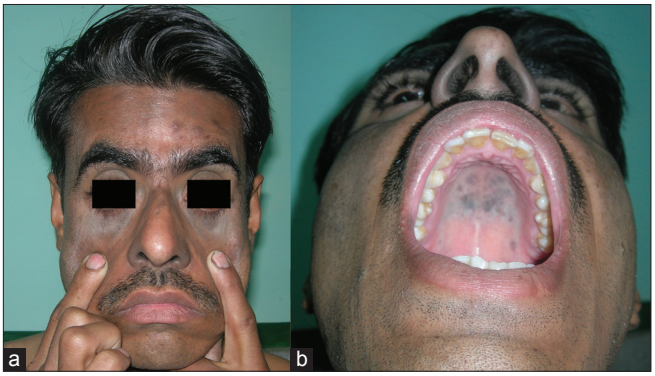


**Figure 3:** (a) Showing Nevus of Ota over the face bilaterally. Note the prominent pigmentation of sclera bilaterally (b) Showing bluish-grey pigmentation over the palate and nasal mucosa bilaterally

**Table 1: Classification of phacomatosis pigmentovascularis[1]**

| Proposed new name | Type of co-existent nevi | Traditional names | Reported additional skin lesions |
|---|---|---|---|
| Phacomatosis cesioflammea | Nevus cesius (blue spot) and nevus flammeus | PPV type IIa/IIb | Nevus anemicus, areas of hairlessness, hypoplastic nails |
| Phacomatosis spilorosea | Nevus spilus (speckled lentiginous nevus) of the macular type and telangiectatic nevus of a pale-pink type | PPV type IIIa/IIIb | Areas of hairlessness, granular cell tumors, lymphedema |
| Phacomatosis cesiomarmorata | Nevus cesius (blue spot) and cutis marmorata telangiectatica congenita | PPV type Va/Vb | None |
| Phacomatosis pigmentovascularis, unclassifiable type | Various types of vascular and pigmented nevi, sometimes with overlapping phacomatosis cesioflammea and phacomatosis spilovascularis | PPV type IVa/IVb and no name | Café au lait macules, hypomelanotic macules, nevus anemicus, nevus sebaceous |

type of all PPVs. Vidaurri-de la cruz *et al*.[6] in a series of 24 consecutive cases of PPV did not find any PPV other than type II.

The pathogenesis of PPV is still controversial. It has been proposed that the combination of vascular and pigmentary anomalies arise as a result of "Twin spotting" phenomenon.[7] Two different recessive mutations could be present on each chromosome of the same pair, on different loci; one each for pigmented and vascular lesions. During embryogenesis, some somatic crossing over could occur with a mitotic recombination, resulting in homozygous cell population in different areas leading to pigmented and vascular nevi.

KTS manifests as a triad of capillary malformations, congenital varicose veins and hypertrophy of underlying tissue. Unilateral lower limbs are frequently involved. KTS may sometimes be seen in association with PPV.[8] The association of phacomatosis cesioflammea with KTS in our patient is indeed interesting and rare.

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