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Case Report

Phacomatosis pigmentovascularis: a rare case report

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ABSTRACT

Phacomatosis pigmentovascularis is a rare group of syndromes characterized by the co-existence of a vascular nevus and a pigmentary nevus with or without extracutaneous systemic involvement. The existing classifications of phacomatosis pigmentovascularis are based on phenotypic characteristics. We report a case of phacomatosis pigmentovascularis, with nevus flammeus, nevus anaemicus and dermal melanocytosis without extracutaneous manifestations. The molecular basis of phacomatosis pigmentovascularis is yet to be elucidated. Whether the various subtypes of phacomatosis pigmentovascularis are separate molecular entities or phenotypic variants of the same disease needs to be settled.

Keywords: Phacomatosis pigmentovascularis, Nevus flammeus, Nevus anaemicus, Dermal melanocytosis

INTRODUCTION

Phacomatosis pigmentovascularis is characterized by the concomitant occurrence of a vascular anomaly, i.e. a capillary malformation (port wine stain) and a pigmentary anomaly in the form of dermal melanocytosis, epidermal nevus, nevus spilus, nevus anemicus and café au lait macules.[1] We report a case of phacomatosis pigmentovascularis with a port wine stain, cherry angioma, dermal melanocytosis, nevus anaemicus.

CASE REPORT

A 22 year-old male presented to the skin outpatient department (OPD) with complaints of a blue-gray discoloration on the left side of the face since birth which was asymptomatic. On dermatological examination, there was a bluish-gray coloured lesion on the left side of the face with bluish discoloration of ipsilateral sclera implying a nevus of Ota (Figure 1). In addition at left clavicular region a hypopigmented patch with serrated margins since birth, which maintain persistent pallor on vigorously rubbing or application of heat or cold (nevus anaemicus) (Figure 2). Over the back there is dermal melanocytosis (giant Mangolian spot) since birth (Figure 3). Also on the left lower region of back he had a Café-au-lait macule (CALM) (Figure 4).

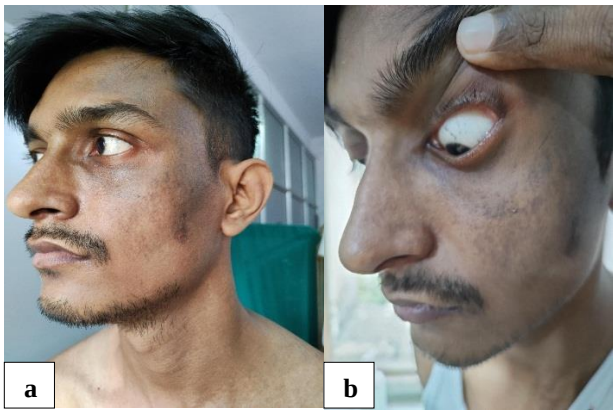

Figure 1: (a) Bluish-gray discoloration on the left side of the face (Nevus of Ota); and (b) ipsilateral bluish discoloration of sclera.

He had sharp, erythematous patches over right anterior forearm and left anterior leg which blanched on pressure

suggestive of a capillary malformation (port wine stain) with overlying multiple dome-shaped, bright, ruby-colored papules (Cherry hemangiomas) (Figure 5). He gave no history suggestive of any central nervous system and other ocular involvement.

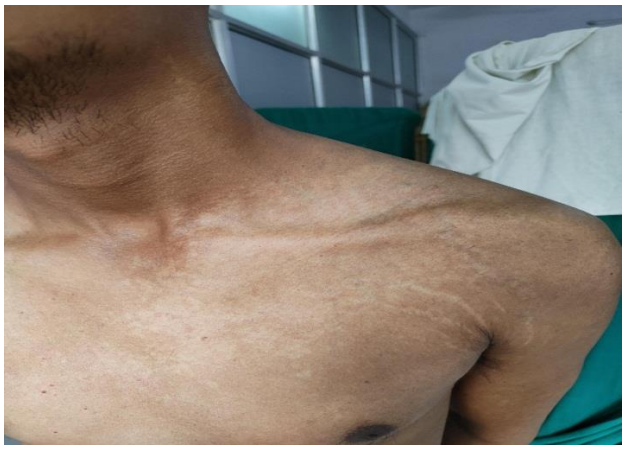

Figure 2: Hypopigmented patch with serrated margins since birth (Nevus anaemicus).

Figure 3: Dermal melanocytosis (giant mangolian spot) since birth.

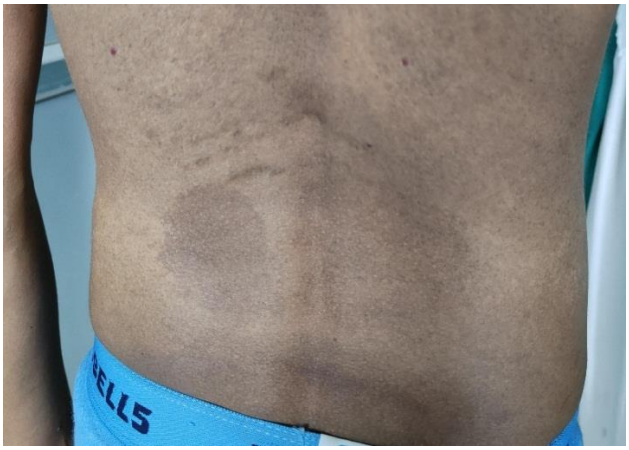

Figure 4: Café-au-lait macule (CALM) at left lower back.

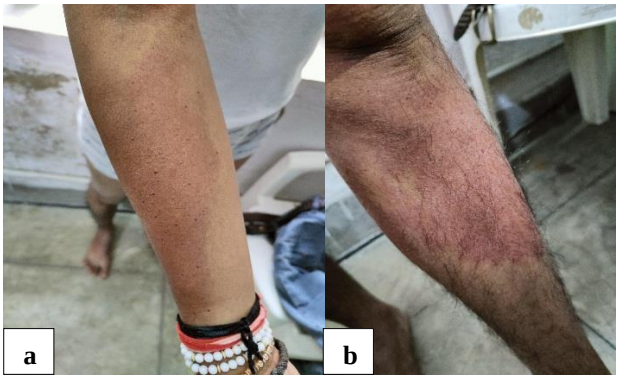

Figure 5: (a) Sharp, erythematous patches (port wine stai) over right anterior forearm and left anterior leg; and (b) with overlying multiple, dome-shaped, bright ruby colored papules (cherry hemangioma).

DISCUSSION

This rare condition was first described by Ota et al in 1947.[2-4] From the clinical features, it was traditionally classified into five types.[2,4,5] Recently, Happle has re-designated this syndrome into four newer types (Table 1).[6] Each division is subdivided into "a" and "b" depending on the presence or absence of systemic involvement respectively. Although there are several sporadic case reports, very few descriptive case series have been published.[3-10]

Port wine stain is one of the commonest vascular malformations usually present at birth and growing in size as the child ages. It is usually associated with hypertrophy of the underlying structures.

The commonest dermal melanocytosis to occur in phacomatosis pigmentovascularis is Mongolian spots, but there have been few reports in literature regarding the concomitant occurrence of nevus of Ota and nevus of Ito with a capillary malformation.[11] Nevus of Ota and Ito are considered aberrant Mongolian spots.[11]

Nevus of Ota also known as nevus fuscoceruleus ophthalmomaxillaris is a dermal melanocytosis which presents from birth with bluish discolouration of unilateral cheek and sclera. It may rarely be bilateral. Nevus of Ito, another form of dermal melanocytosis goes by the name, nevus fuscoceruleus acromiodeltoideus. It presents as brown to bluish gray pigmentation over the shoulders. When suspecting a case of phacomatosis pigmentovascularis, ocular and neurological abnormalities will have to be ruled out.

Ocular complications include ocular melanosis, glaucoma, iris hamartomas, mammillations and nodules.[12] Neurological involvement is in the form of delayed development, seizures, hydrocephalus, intracranial calcifications and cerebral atrophy.[13]

Prognosis of the disease depends on whether systemic involvement is present or not. Treatment of cutaneous lesions is for aesthetic purposes and includes Q switched alexandrite laser for the pigmentary lesions and pulsed dye laser for the vascular lesions.[14]

Table 1: Classification of PPV.[6]

Hasegawa's classification	Happle's classification	Vascular anomaly	Pigmentary anomaly
I	Non-existent	Nevus flammeus	Epidermal nevus
II	Phacomatosis cesio flammea	Nevus flammeus+nevus anaemicus	Dermal melanocytosis
III	Phacomatosis spilorosea	Nevus flammeus+nevus anaemicus	Nevus spilus
IV	Unclassified	Nevus flammeus+nevus anaemicus	Dermal melanocytosis+nevus spilus
V	Phacomatosis cesio marmorata	Cutis marmorata telangiectatica congenita (CMTC)	Dermal melanocytosis

CONCLUSION

Our case can be classified as phacomatosis pigmentovascularis type IIb.

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