

## Phacomatosis pigmentovascularis Type IIa – A case report

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### ABSTRACT

Phacomatosis pigmentovascularis (PPV) is a rare syndrome characterized by capillary vascular malformation and pigmented nevus but with a wide variability in clinical presentation. A case of a 9-month-old patient is reported, who presented with capillary malformation and hemihypertrophy. These features typically are seen in Klippel–Trenaunay (KT) syndrome, a syndromic type of congenital vascular malformation. However, in addition, this child had large persistent Mongolian spots, the presence of which allowed us to classify this case as PPV, type II. This is a condition with a clinical course not always benign as KT syndrome.

**Key words:** *Mongolian spots, Nevus flammeus, Vascular malformations*

Capillary vascular malformations are not uncommon, and when they occur in isolation, they may cause significant psychosocial disability but without any increase in mortality or morbidity. However, when they occur with dermatologic anomalies, they constitute a rare syndrome called as phacomatosis pigmentovascularis (PPV) [1] and mandate a thorough workup to rule out significant neurologic and ocular involvement. Around 245 cases have been published since it was described in 1947 [1]. The present case is reported because we believe that though most clinicians would be familiar with the better-known syndromes of capillary malformations, awareness about the significance of its association with pigmented lesions may be lacking.

### CASE REPORT

A 9-month-old female, the first issue of non-consanguineous marriage, presented with a capillary vascular malformation involving the entire right upper limb. Face and other parts of the body were not involved. The lesions were present since birth, and neither regressed nor enlarged with time. The child was otherwise asymptomatic. There was hypertrophy of the right upper limb (Fig. 1) with no significant limb length discrepancy. In addition, there were large Mongolian spots which were present since birth and covered almost the entire back (Fig. 2) and part of the chest. There was no developmental delay or seizures. Neonatal period was uneventful and past history was non-contributory. No abnormal findings were detected on systemic examination.

Imaging studies did not reveal any evidence of arteriovenous (AV) malformation or intracranial lesions, and there was no

evidence of ocular involvement in the form of melanosis bulbi or glaucoma. There was no clinical or radiological evidence of varicosity of veins. Our initial diagnosis was Klippel–Trenaunay (KT) syndrome, and we had presumed that the Mongolian spots were normal for age; though they were slightly larger in size than expected for the age. Parkes Weber, Sturge–Weber, and Proteus syndrome were also considered based on the presenting features of vascular malformation and hemihypertrophy. After review of the literature on the presenting features along with persistent Mongolian spots, we made the diagnosis of phacomatosis pigmentovascularis type II. Parents were advised regular follow up to look for systemic involvement.

### DISCUSSION

This clinical picture would invariably prompt a diagnosis of KT syndrome, which is also a rare condition and presents with capillary malformation on an extremity and hypertrophy of the involved limb. The distinguishing feature is that the vascular lesion in KT is not accompanied by pigmented nevus or Mongolian spots as in our case. Parkes Weber syndrome also presents with vascular malformation and limb hypertrophy but is differentiated by the presence of multiple, small, fast flow AV shunts detected by ultrasound and magnetic resonance imaging.

Presence of pigmented nevus in addition to the vascular malformation and limb overgrowth distinguishes the condition as phacomatosis pigmentovascularis which is an extremely uncommon, sporadic, genetic condition with no documented sex predilection. This condition was described first by Ota in 1947 [1] and till date only 245 cases have been reported [2]. This association has been categorized into following five types



**Figure 1: Capillary malformation, hypertrophy of the right upper limb**



**Figure 2: Mongolian spots covering most of the back**

by Hasegawa and Yasuhara based on pigmentary lesions, (I) epidermal nevus, (II) Mongolian spots, (III) nevus spilus, (IV) nevus anemicus, and (V) marmorata telangiectatica [3,4], and two subtypes “a” (cutaneous only manifestations) and “b” (associated with systemic manifestations including ocular, vascular, skeletal, and neurological) [2]. Subsequently, Happle in 2005 proposed a new simplified classification into three groups based on the skin lesions – (I) Mongolian spots,

(II) nevus spilus, and (III) cutis marmorata telangiectatica congenital [5]. Our case fits into type IIa PPV or cesioflammea.

The diagnosis is clinical, and investigations are proposed for the purpose of classification. The prognosis of PPV is determined by the nature and extent of systemic involvement which may manifest over a period of time [6].

## CONCLUSION

PPV is a rare case but it is important that clinicians are aware of this condition for two reasons: (a) presence of a dermatologic lesion with a vascular malformation mandates a thorough workup for systemic involvement as it may alter the clinical course, (b) close follow-up of all children must be advised even in the absence of systemic involvement at the time of presentation.

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