

A Rare Case of Phakomatosis pigmentovascularis

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Abstract: Phakomatosis pigmentovascularis (PPV) is a rare syndrome characterized by extensive vascular naevus with pigmentary naevus with or without systemic manifestations. PPV has been classified into 4 types with subtype "a" for cutaneous involvement only and subtype "b" for cutaneous and systemic involvement. A case of 52 year old is reported, who presented with extensive naevus flammeus and verrucous epidermal naevus with episodes of seizure as systemic complain. This manifestation allows us to classify this as a case of Type 1b Phakomatosis pigmentovascularis, according to Hasegawa's classification. This case is presented due to the rare nature of the disease.

Keywords: Phakomatosis Pigmentovascularis.

INTRODUCTION

First reported in 1947, it is a rare congenital nevoid syndrome. Most case reports originate in Japan. The major clinical manifestation consists of coexisting extensive nevus flammeus and pigmentary nevus with or without systemic involvement. Abberantmongolion spots, nevus anemicus, pyogenic granulomas, Becker nevus, iris mammillations, renal angiomas renal agenesis and moyamoya disease have been reported as associations [1-4]. Patients with phakomatosis pigmentovascularis can present with clinical manifestations of one or more of the following: Sturge-Weber syndrome, Klippel-Trenaunay syndrome and melanosis oculi.

Four distinct subtypes have been reported: type 1(port-wine stain and linear epidermal nevus), type 2(port-wine stain and dermal melanocytosis), type 3(port-wine stain and nevus spilus) and type 4(port-wine stain, dermal melanocytosis and nevus spilus). Type 2, 3 and 4 can also have associated nevus anemicus. Two different lasers have been used to treat the vascular and pigmentary lesions in the same patients: Q-switched ruby laser irradiation to the site of dermal melanosis and pulsed dye laser irradiation to the port-wine stain [5].

CASE HISTORY

- A 52-year male presented with complains of
- Reddish-pink macular lesions on both sides of face since birth
 - Overlapping dark nodular masses over face also since birth

The lesions were asymptomatic

Past History: There was ant past history of seizures (2-3times) for which he was taking anti-epileptic medication.

Family History: No relevant family history.

Personal History: Nothing significant, Diet – mixed

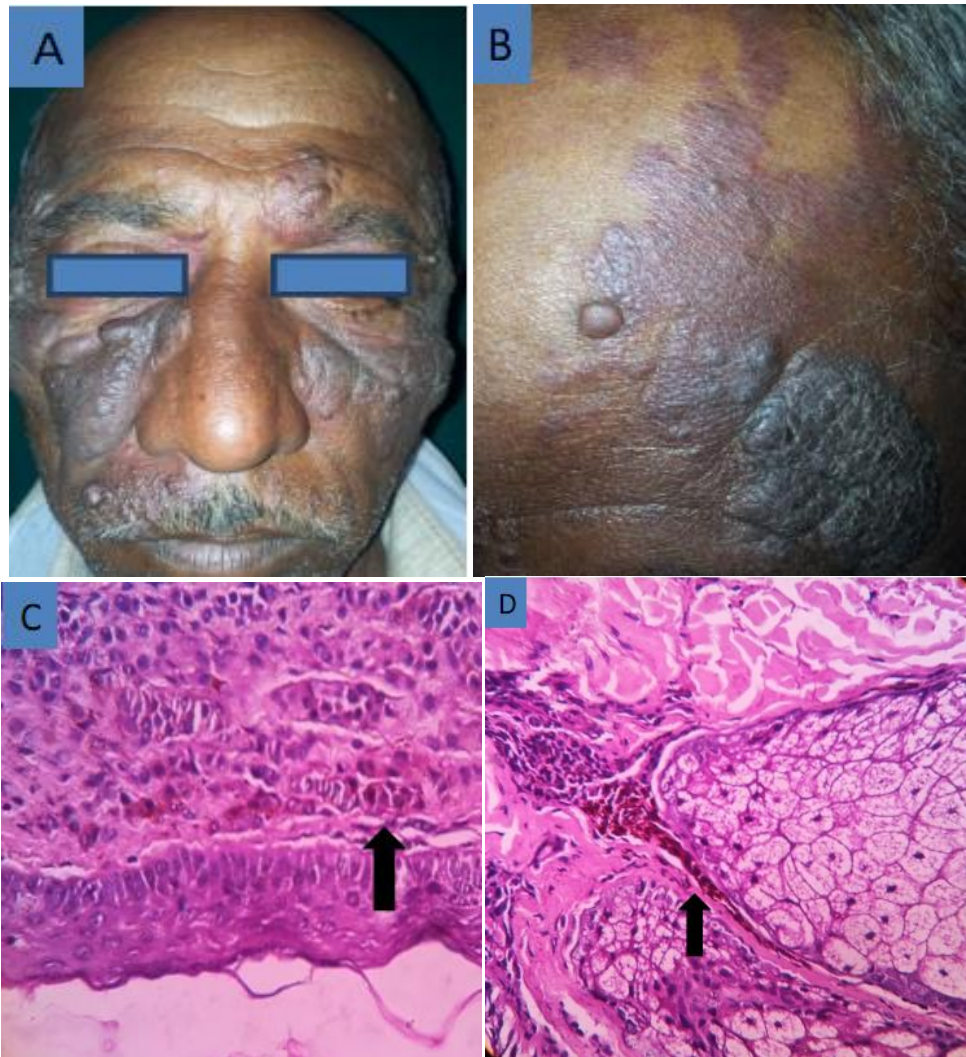
Clinical examinations

On physical examination, his blood pressure was 110/70 mmHg and her pulse was 72/minute. . No pallor, Icterus, oedema feet was noted, of lower thoracic spine was seen. RS, CVS, P/A & CNS Examination was normal. Cutaneous examination revealed nevus flammeus with overlapping dermal melanocytic nevus on bilateral malar prominences' and scalp, Neurologic and ophthalmic examination was normal, Limb circumferences were normal with no evidence of varicose veins.

Investigations

Hb-13.4gm/dl, TLC- 7500/cumm, MCV 74
Liver Function: within normal limit
Kidney Function: within normal limit
ESR - 05mm (normal range female: 0-22 mm)
Urine microscopy: within normal limit
Skin biopsy: On histopathological examination the presence of dermal melanocytosis with capillary venous malformation was seen (nevus flammeus)

And patient was referred to neurosurgeon for biopsy and management.



**Fig-A and B: Nevus flammeus and dermal melanocytic naevus.
Fig-C and D: Dermal melanocytic cell nests**

DISCUSSION

Phakomatosis pigmentovascularis (PPV) is thought to be an embryogenic anomaly affecting the vasomotor nerves and the melanocytes, both derived from neural crest. Clinically, PPV manifests as a large capillary malformation, which is metameric in distribution and mainly located on the trunk or the extremities, in association with pigmented cutaneous lesions, such as a pigmented nevus, a nevus spilus, a café au lait patch, or an atypical Mongolian spot [6]. That is not located on the sacrum Nevus anemicus [8] can also be seen in the vicinity as a twin spot [7]. These cutaneous lesions can be associated with systemic, visceral (e.g., larynx hypoplasia, intestinal polyposis), muscular (scoliosis), neurologic (mental retardation, epilepsy, intracranial calcification, cerebral atrophy), or ocular involvement (anomaly of the iris).

The term phakomatosis refers to a developmental malformation simultaneously affecting the eye, skin and central nervous system.

Classification of phakomatosis pigmentovascularis:

Subtype A: Cutaneous forms

Type 1A: port-wine stain/capillary malformation (CM) □□epidermal naevus Type 2A: CM □□Mongolian blue spots □□naevus anaemicus Type 3A: CM □□naevus spilus□□naevus anaemicus Type 4A: CM □□naevus spilus□□naevus anaemicus□□aberrant Mongolian blue spots

Subtype B: Systemic/syndromic form Types 1–4 B as above plus Sturge–Weber syndrome or naevus of Ota or Klippel–Trenaunay syndrome

The classification of this disorder has been

simplified; it has been reported that cutis marmorata and aberrant Mongolian blue spots define a fifth type. The condition is uncommon and arises sporadically. It has been hypothesized that twin spots account for this phenotype. In this hypothesis, somatic mutations on nearby genes lead to mosaic spots in close proximity to one another. Type 2 is reported to account for 80% of all cases. The capillary malformation may occur anywhere on the body, and can be segmental. The aberrant Mongolian blue spots occur on atypical sites and tend to persist, unlike conventional

Mongolian blue spots. Affected cases should be investigated depending upon the sites involved, for example, for glaucoma if the CM affects the eyelids or for KTS if there is limb involvement

CONCLUSION

- Phacomatosis pigmentovascularis is a rare disorder, which is usually benign.
- Association with a neuro cutaneous disorder must be kept in mind and CT scan should be done when presenting with systemic symptoms (like seizure in our case).
- To the best of our knowledge, this is the first case in India of PPV type IIb presenting in old age (52yr) with tumor of the base of the skull.

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