

# PHAKOMATOSIS PIGMENTOVASCULARIS: REPORT OF A CASE

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*Abstract—The present study introduces a rare case of phakomatosis pigmentovascularis, which is characterized by the existence of pigmentary naevus and vascular naevus. Until 1985, 63 cases of this syndrome have been reported, mostly in Japan (56 cases). This is the first case of phakomatosis pigmentovascularis, reported in Iran.*

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*Key words: phakomatosis pigmentovascularis; portwine stain (naevus flammeus); Mongolian spot; naevus spilus; naevus anaemicus*

## INTRODUCTION

The word phakomatosis implies the developmental malformations involving skin, eye and central nervous system (CNS) simultaneously.

Phakomatosis pigmentovascularis is a syndrome which consists of vascular naevi of portwine stain type, oculocutaneous melanosis with or without CNS manifestations such as fits and hemiplegia (1,2,3). This syndrome has much in common with Sturge-Weber syndrome, but it may only consist of cutaneous manifestations (1,2,3).

According to the suggested classification (2), the case reported in the present study, is termed phakomatosis pigmentovascularis type IV-a.

## CASE REPORT

An 8-year-old Iranian girl was referred to the Out-patient Department of Razi Hospital in December 1993 with the chief complaint of diffuse skin discoloration since birth.

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She was the product of a full-term normal vaginal delivery. Her parents were cousins. The patients had normal intelligence, and had no history of seizure. No other member of her family had any skin lesion similar to those of the patient.

She was a relatively well-developed, well-nourished girl. Physical examination of skin showed three kinds of naevoid lesions over various parts of the body; these included portwine stains, aberrant Mongolian spots and a brown macule of naevus spilus. The portwine stain characterized by a red, reticulate macular patch, was present on the left side of the face, over the cheek and lower lid, extending from the left ear to the left side of the neck. There were the same type of lesions on the right lower lid, right infra-orbital area, and both temples (Fig. 1). Several scattered patches of similar but paler lesions with irregular borders were present on both upper and lower limbs and back of the hands (Fig. 2).

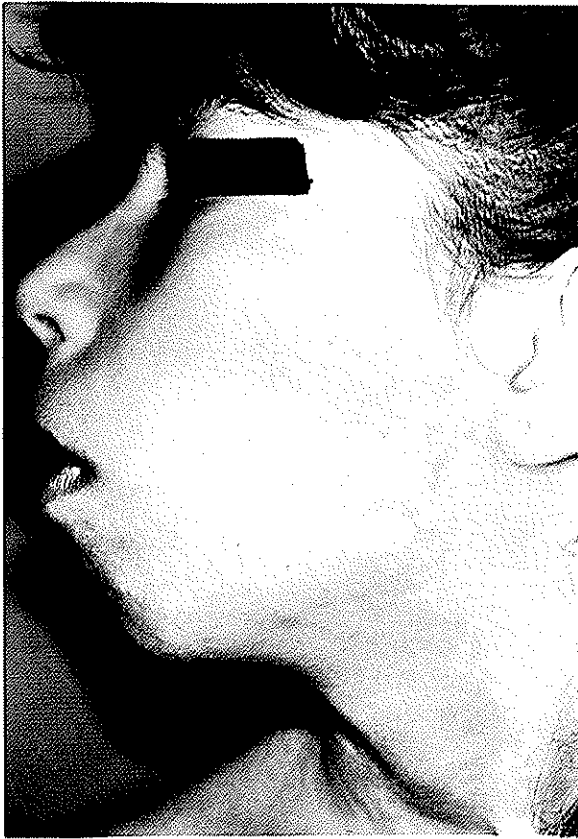
The aberrant Mongolian spots as large, irregular, pale, and blue patches were present on the chest, back and thighs, overlapped by the above-mentioned scattered portwine lesions (Fig. 2).

The lesion of naevus spilus (speckled and lentiginous naevus) included a 1×0.5-cm light brown patch with dark brown speckled areas on it, which was present on left scapula since birth (Fig. 3).

There were several small blue patches in the sclerae of both eyes (melanosis bulbi). Palpebral conjunctivae were not discolored (Fig. 4).

In the left side of oral mucosa (cheek and palate) several pale and red patches of portwine type were present.

There was no discrepancy in length and size of the limbs. The general and neurologic physical examination revealed normal findings (confirmed by neurologist). Ophthalmologic examination showed myopia and pigmentary and vascular changes at middle and inner coats of the eyes. Intraocular pressure was normal.



**Fig. 1.** The patches of portwine stain on the face and neck (lateral view).



**Fig. 3.** Speckled and lentiginous naevus (naevus spilus) on the back.



**Fig. 2.** The patches of portwine stain, superimposed by Mongolian spots, on the chest and upper limbs.



Fig. 4. Blue lesions in the sclerae of both eyes (melanosis bulbi).

Paraclinical studies (complete cell blood count, liver function test, urine analysis, BUN, CT scan and chest x-ray) were all within the normal limits.

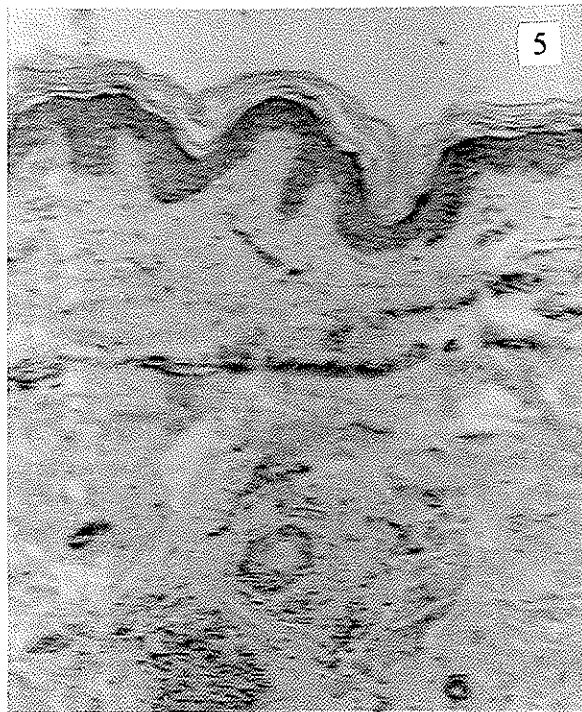
Histopathological examination of the blue spots showed a few fusiform melanocytes with melanin granules in the dermis, compatible with the diagnosis of Mongolian spot (Figs. 5 and 6). Biopsy specimen of the brown patch showed elongation of rete ridges along with hyperpigmentation of the basal layer, nests of naevus cells, and macrophages containing melanin within the dermis. These findings were in favor of the diagnosis of naevus spilus (Fig. 7).

## COMMENT

Phakomatosis pigmentovascularis was first reported in Japan in 1947 (2). More than 63 cases were reported from 1947 till 1985, mostly in Japan (59 cases) (2). Some of these cases had systemic involvement and some had not (the localized type) (2). The systemic involvement included CNS and ocular signs and symptoms of Sturge-Weber syndrome, malignant colon polyposis (3), hemihypertrophy of Klippel-Trenaunay syndrome, and etc (2). The accepted classification of phakomatosis pigmentovascularis, is shown in Table 1.

Table 1. Classification of phakomatosis pigmentovascularis.

Type	No. of case reported in the literature	Pattern of pigmentary and vascular naevi
Ia (localized)	3	portwine stain and naevus
Ib (systemic)	0	pigmentosus et verrucosus
IIa (localized)	26	portwine stain and blue spots
IIb (systemic)	25	with or without naevus anaemicus
IIIa (localized)	3	Portwine stain and naevus spilus
IIIb (systemic)	0	with or without naevus anaemicus
IVa (localized)	3	portwine stain, blue spots and naevus
IVb (systemic)	3	spilus, with or without naevus anaemicus



Figs. 5 and 6. Fusiform melanocytes with melanin granules in the superficial and deep dermis (Mongolian spots).

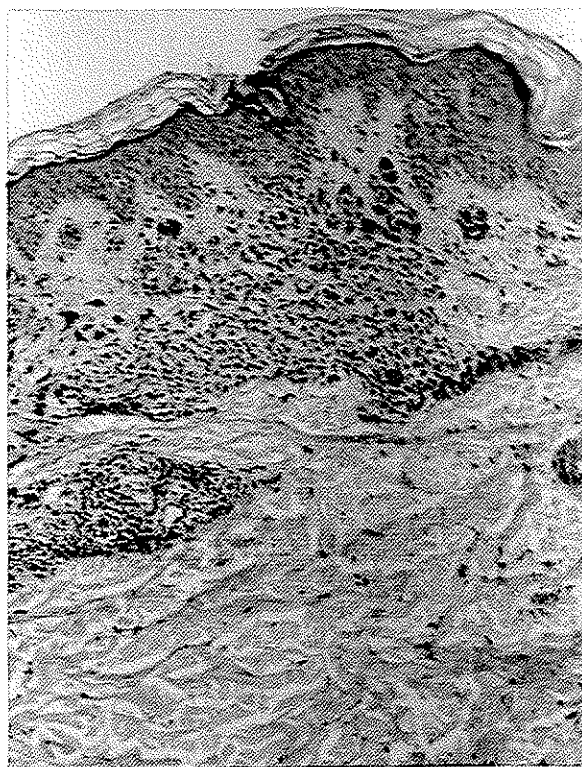


Fig. 7. Elongated rete ridges, diffuse hyperpigmentation of basal layer and nests of naevus cells in the dermis (naevus spilus).

According to this classification the case we have reported in this study, is termed phakomatosis pigmentovascularis type IV-a because the patient had no evidence of systemic involvement other than oculocutaneous discoloration changes.

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