Phakomatosis Pigmentovascularis

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Phakomatosis pigmentovascularis was first described in 1947 as a distinctive association of cutaneous hemangioma and pigmentary nevi by Ota et al.

We describe a 7-year-old boy who, since birth, had three kinds of discolored patches over the various parts of the body: blue spots (dermal melanocytic nevi), reticulated reddish patches (nevus flammeus), and hypopigmented macules (nevus anemicus). No systemic disease was found. (Ann Dermatol 4:(2) 103–107, 1992)

Key Words: Dermal melanocytic nevi, Nevus anemicus, Nevus flammeus, Phakomatosis Pigmentovascularis

Phakomatosis pigmentovascularis (PPV) was first described in 1947 as a distinctive association of cutaneous hemangioma and pigmentary nevi[1]. This disorder has been reported mostly in Japan. In Korea, the term PPV was first reported in 1985 by Youn et al[2]. Nine cases of this disorder have been reported in Korean literature.

We report a classic case of PPV, type II_a with review of the literature.

REPORT OF A CASE

A 7-year-old boy visited our department because of extensive red and gray-blue cutaneous pigmentations since birth.

He was the product of a full-term normal delivery. No medication was taken by the patient's mother during pregnancy. He was born with an extensive nevus flammeus involving the face and extremities. There were bluish stains on the trunk and both shoulders. Infantile growth and development were normal. Bluish macules on both sclerae were noted at 4 years of age.

Physical examination revealed three kinds of discolored patches over the various parts of the body. Extensive diffuse gray-bluish pigmented patches were found on both shoulders, chest, abdomen, back, buttocks and extremities (Fig. 1.2). Just as Mongolian spots, they were completely flat and had no infiltration. Both sclerae and the hard palate were also pigmented (Fig. 3.4). Reticulated telangiectatic erythematous patches suggesting nevus flammeus were located on the chest, abdomen, back, buttocks and extremities (Fig. 1.2). The other kind of the discolored patches, hypopigmented macules suggesting nevus anemicus, were located on the buttocks and extremities. These hypopigmented macules could not be made red by stroking (Fig. 5). There was no consanguinity in his parents. His sister had blue macules on both sclerae. His intelligence and physical constitution were normal. There were no discrepancies in leg and arm lengths or sizes, and the percentiles of height and weight were normal.

Laboratory studies, including complete blood cell count, urine analysis, liver function test, renal function test, stool examination and VDRL were within normal limits. ECG and EEG were normal. X-ray films showed no abnormalities in the chest, upper gastrointestinal tract, colon and skull. Computed tomographic scan of the brain also showed no abnormalities. Otolaryngologic and ophthalmologic examinations were essentially normal.

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This case was presented at the 43th Annual Spring Meeting of the Korean Dermatologic Association of April 19, 1991 at Jeonju, Korea.

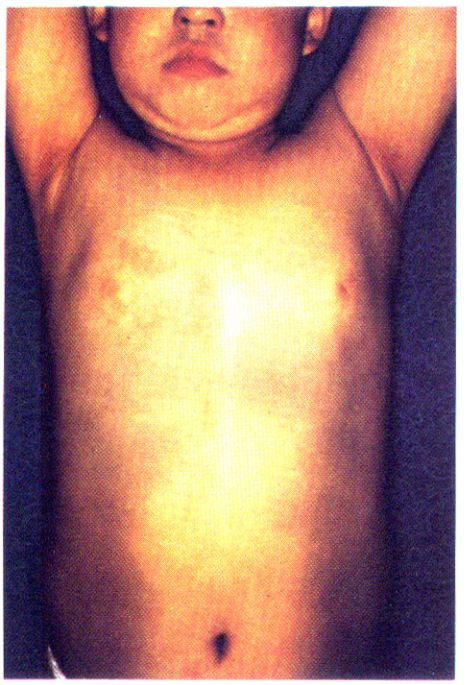

Fig. 1. Nevus flammeus and gray-bluish pigmentation on the upper arms, thorax and abdomen.

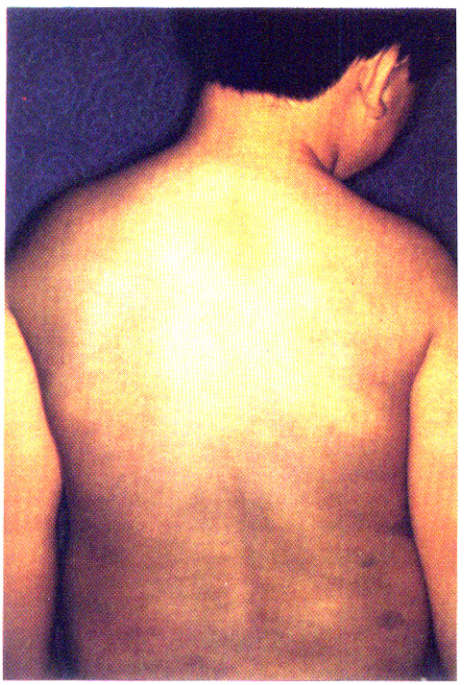

Fig. 2. Nevus flammeus and gray-bluish pigmentation on the upper arms, shoulder and back.

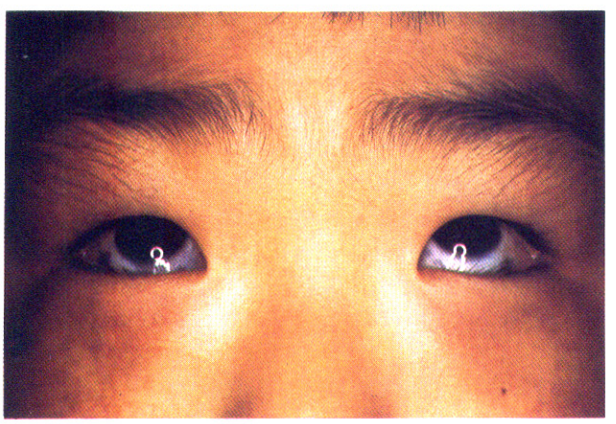

Fig. 3. Melanosis oculi on both sclerae.

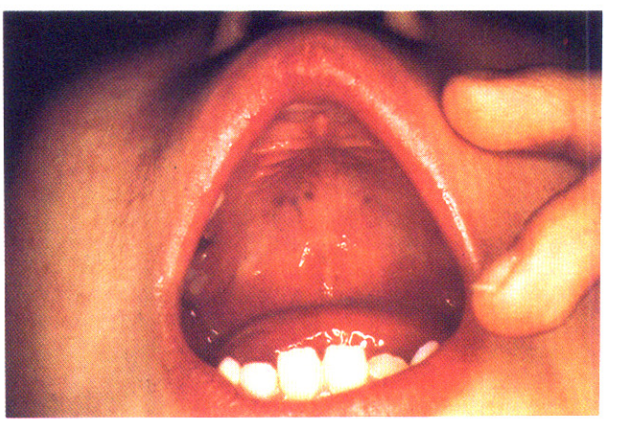

Fig. 4. Symmetric bluish spots on hard palate.

Histopathological examination of the nevus flammeus in the left forearm demonstrated mild dilatations of the capillaries in the upper dermis and mild perivascular infiltration (Fig. 6). A biopsy specimen of the blue spots in the left shoulder showed a few spindle shaped melanocytes with melanin granules scattered among the collagen bundles in the upper and mid dermis. These cells showed dark brown stained melanin granules in the Fontana-Masson stain (Fig. 7).

He was not given any treatment. When and if functional disturbances appear, he will be treated.

DISCUSSION

The term phakomatosis was coined in 1920 by van der Hoeve to describe conditions that share characteristic central nerve system and retinal tumors (phakomas)[3]. In 1947, phakomatosis pigmentovascularis (PPV) was first described as a distinctive association of cutaneous hemangioma and pigmentary nevus by Ota et al[1]. The disorder was subclassified into two types: type I, or the Adamson-Best type, and type II, or the Takano-Krüger-Doi type. The former consisted of nevus

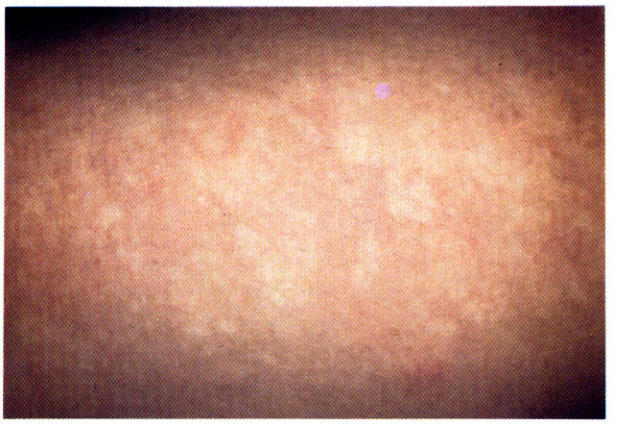

Fig. 5. Nevus anemicus, located on the extremities, did not develop a flare after rubbing of the skin.

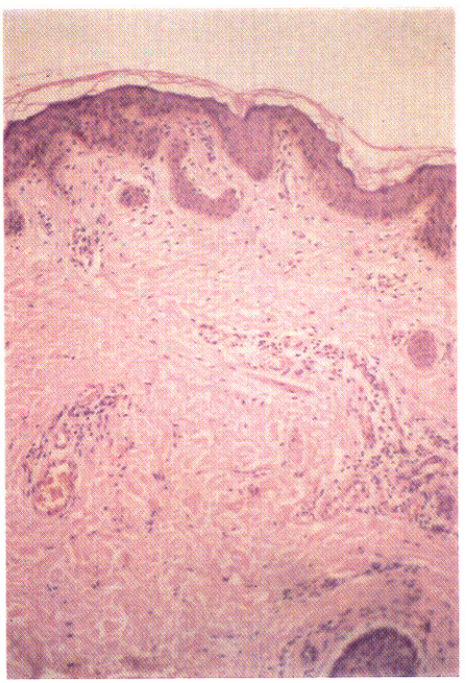

Fig. 6. This biopsy specimen taken from the reddish patch on the right forearm shows mild dilatation of the capillaries and perivascular inflammatory infiltrates in the dermis (H & E stain, ×100).

flammeus and nevus pigmentosus et verrucosus, and the latter of nevus flammeus and aberrant mongolian spots[4-6]. In 1966, Toda reported a case of coexisting nevus flammeus and nevus spilus as the Kobori-Toda type, now classified as type III[7]. In 1979, Hasegawa and Yasuhara proposed a classification of PPV with four subdivisions, and each of them which could be localized or systemic (Table 1)[8]. According to Tamotsu et al[9], among

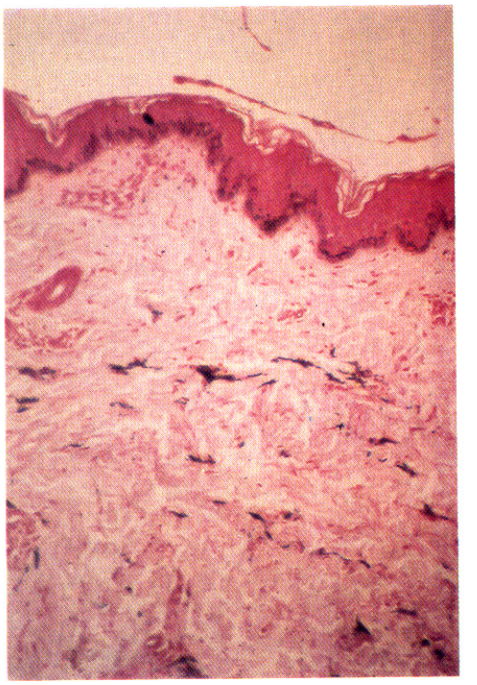

Fig. 7. A biopsy specimen taken from the blue spots on the left shoulder shows black-colored melanin in the dermal melanocytes (Fontana-Masson stain, ×100).

seventy-four cases of PPV reported in Japan, sixty-three cases were type II. Type I has never been found. And only four cases were type III. In our case, nevus flammeus, blue spots, and nevus anemicus were found concurrently. No systemic disease of any kind has been found. According to Hasegawa's classification, our case is classified as type IIa.

In Korean literature, Kim et al[10] reported two cases of bilateral Ota nevus and bilateral Ito nevus in 1981. One of them had a diffuse bluish pigmented patches on the nearly whole body, reticulated patches of nevus anemicus on the dorsal back and both arms and nevus flammeus on the right cheek. We think this case as the first report of PPV in Korea although the authors did not used the term of PPV. The term of PPV was first reported in 1985 by Youn et al[2]. Since then, four papers on PPV have been published[11-14]. These ten cases of PPV, including our case, are classified and categorized in Table 2. Only the second type is similar to the original description of Ota and to this case.

In systemic type of PPV, abnormalities in organs other than skin have been reported. In Hasega-

Table 1. Classification of phakomatosis pigmentovascularis (Hasegawa and Yasuhara)

Type		Pattern of pigmentary nevus and vascular nevus
I	Ia (localized) Ib (systemic)	Nevus flammeus and nevus pigmentosus et verrucosus
II	IIa (localized)	Nevus flammeus and blue spots, with or without nevus anemicus
III	III (localized)	Nevus flammeus and nevus spilus, with or without nevus anemicus
IV	IVa (localized)	Nevus flammeus, blue spots, and nevus spilus, with or without nevus anemicus

Table 2. Summary of reported cases of phakomatosis pigmentovascularis in Korea.

Author, year	Patient Age, yr/Sex	Type	Nevus anemicus	Systemic disease
Kim et al[10], 1981	15/F	IIb	+	−
Youn et al[2], 1985	2/F	IIb	−	Soft tissue hypertrophy
	1/F	IIb	−	VSD, abnormal EEG
	2/M	IIb	−	Sturge-Weber syndrome
	5/M	IVb	−	Abnormal EEG
Lee et al[11], 1989	23/F	IIb	−	Congenital glaucoma, cataract
Choi et al[12], 1990	2/F	IIa	−	−
Kim et al[13], 1990	4/F	IIa	−	−
Choi et al[14], 1991	12/M	IIa	−	−*
Our case	7/M	IIa	+	−

*: Complicated by congenital temporal alopecia

wa and Yasuhara's review of 63 cases (28 cases of systemic type), 14 were associated with Klippel-Weber syndrome and 17 with Sturge-Weber syndrome[15]. The other reported systemic complications are malignant colon polyposis, scoliosis, leg-length discrepancy, anemia, ocular alterations (glaucoma and buphthalmos), mental disturbance and seizures. In our case, we also examined systemically, but we could not find any other associated anomalies. Follow-up should be continued periodically to search for other functional disturbances.

The pathogenesis of this complex malformation remains unclear. In 1977, Kawamura stated that the pathogenesis of the phakomatoses must lie at the germ level, but he failed to clarify which germ cells produced the phakomatoblasts of PPV[16]. Ruiz-Maldonado et al stated that the hypothetical pathogenic factors (drugs, virus, substances toxic to the nervous system, etc.) could have an irritating effect and cause some clones of angioblasts and melanoblasts to proliferate in an aberrant form[17]. In 1985, Hasegawa and Yasuhara concluded that all pigmentary and vascular nevi were caused by functional disorders of vasomotor nerve cells and abnormal melanocytes, which originated in the embryonal neural crest[15]. In cases associated with systemic diseases, many of the involved sites can be attributed to this organ of cell origin, since some cases show lesions that have developed from cells derived from the embryonal mesenchyme[15].

The genetic features of PPV are unclear. In this case, the patient's sister had blue macules on both sclerae, but his parents had no skin lesions similar to those of the patient. Most of reported cases

were sporadic, but some cases had familial background[15-18]. This family history suggests that hereditary factor may also play a role in the manifestation of PPV.

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