Phakomatosis pigmentovascularis type IIa

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Phakomatosis Pigmentovascularis (PPV) is a rare cutaneous congenital malformation syndrome, defined as simultaneous occurrence of congenital cutaneous vascular and pigmentary anomalies. As most of the reported cases are from Far East countries, especially Japan, we reported a case of PPV type IIa in an Iranian patient. This case was unusual because of the coexistence of unilateral extensive port-wine stains all over the left body side and left hard palate and a right plantar port-wine stain.

Keywords: nevus of Ota, phakomatosis pigmentovascularis, pigmentary anomalies, port-wine stains, vascular anomalies

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INTRODUCTION

Today, the term phakomatosis is mainly applied to genetically determined diseases characterized by the presence of two or more different nevi, such as phakomatosis pigmentovascularis or phakomatosis pigmentokeratotica [1].

Phakomatosis Pigmentovascularis (PPV) is a rare cutaneous congenital malformation syndrome, defined as simultaneous occurrence of congenital cutaneous vascular and pigmentary anomalies [2]. There are several associated vascular abnormalities in PPV, but oral involvement has not been reported with this disorder. More than 222 patients with PPV have been reported in the literature, most of whom are sporadic and from Japan, Mexico or Argentina [3]. Herein, we report a patient with a Mongolian spot, nevus of Ota, nevus anemicus and widespread port-wine stains along with oral mucosal involvement.

CASE REPORT

An 18-year-old man presented with unilateral extensive port-wine stains all over his left body side (face, neck, trunk, and lower extremities). He also had a similar red patch on the right plantar surface .In addition, several poorly defined areas showing a diffuse bluish-grey pigmentation were observed on his left shoulder and upper back (Figure 1). He also had a nevus anemicus on his left back side in the vicinity of the port-wine stain. A thorough ophthalmological examination disclosed blue-grey discoloration in the right periorbital

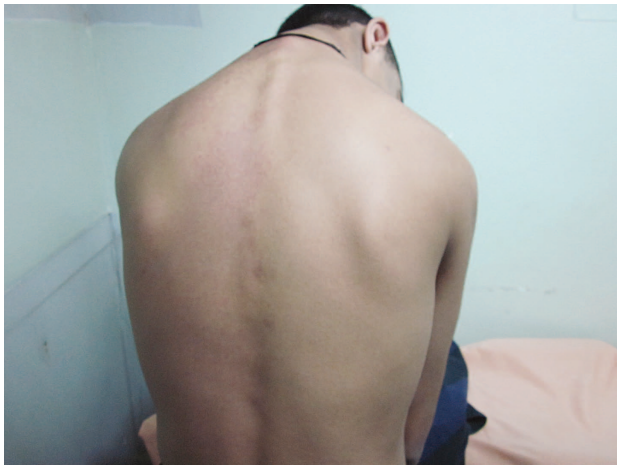

Figure 1. Diffuse bluish-grey pigmentation were observed on his left shoulder and upper back

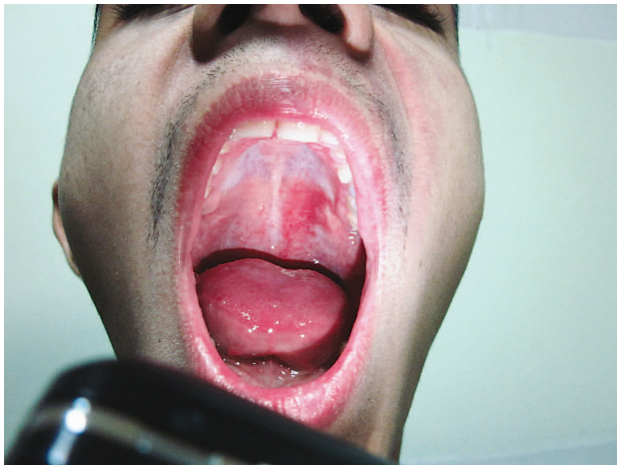

Figure 3. Nevus flammeus was found on the left hard palate

area associated with bluish discoloration of the ipsilateral sclera, a finding consistent with the nevus of Ota (Figure 2). On further examination, a nevus flammeus was found on his left hard palate (Figure 3). His parents were not consanguineous and there was no family history of similar or other skin disorders. No other abnormalities were observed on general physical and neurological examination. Thus, he did not have Sturge-Weber syndrome and had not taken anticonvulsants. He also had normal mental and physical development. Based on the clinical findings, a diagnosis of PPV type IIa was made.

DISCUSSION

PPV was first described by Ota et al in 1947 [2]. In the past, this entity was classified into four types according to the main pigmented lesion [4-6]. Among the known subtypes of PPV, type II is the most common form (80%). Types I, III, and IV are rare and only a few cases of each type are reported in the literature [7]. This classification is as follows:

Type I=nevus flammeus + pigmented linear epidermal nevus

Type II=Nevus flammeus + blue spots ± nevus anemicus

Type III=nevus flammeus + nevus spilus ± nevus anemicus

Type IV=nevus flammeus + blue spots + nevus spilus ± nevus anemicus [8]

Type V=cutis marmorata telangiectatica congenita and blue spots [9].

Each type has been further classified into two subdivisions: (a) cutaneous involvement only, and (b) cutaneous and systemic involvement.

Recently, Happle proposed a new simpler classification for PPVs which divides PPVs into four groups;

Phakomatosis cesioflammea: this type is characterized by coexistence of one or more aberrant blue spots and one or more port-wine stains. The pigmentary nevus of phakomatosis cesioflammea can correspond to a nevus of Ota, a nevus of Ito, or a Mongolian spot.

Phakomatosis spilorosea: it is characterized by the coexistence of nevus spilus of a macular type and a pale-pink nevus (unlike the purple hue of the nevus flammeus). Another type of associated pigmentary spot in the subtype of phakomatosis spilorosea is nevus spilus.

Phakomatosis cesiomarmorata: This type is

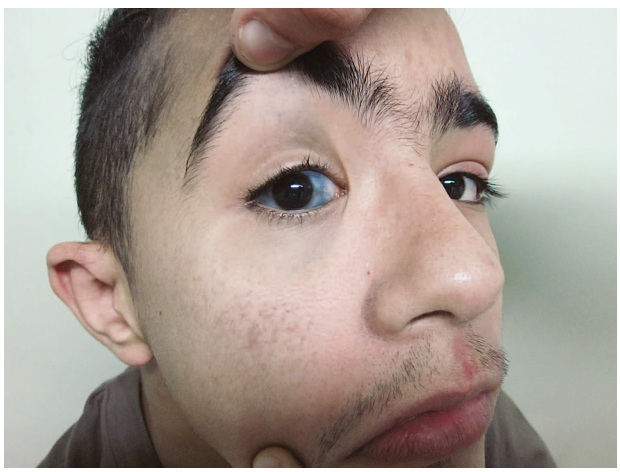

Figure 2. Bluish discoloration of the ipsilateral sclera, a finding consistent with nevus of Ota

characterized by the association of a blue spot with cutis marmorata telangiectatica.

The fourth group is comprised of unclassifiable PPV. In addition, within the unclassified phakomatosis, café-au-lait spots have been reported and are generally large in size [10].

The vascular component of PPV may manifest itself as paired patches of port-wine stain and nevus anemicus. Phakomatosis cesioflammea is always associated with nevus flammeus, which is why nevus roseus has been proposed to represent a distinct type of vascular nevus [11]. It shows a pale pink colour similar to that of a salmon patch throughout life.

Nevus anemicus is sometimes associated with port-wine stains in types II, III and IV. Clinically, a nevus anemicus is a hypopigmented patch similar to vitiligo. However, it is not a disease of the pigmentary system, but rather a localized physiological state of permanent vasoconstriction.

While the pigmentary component may manifest itself as paired patches of hyperpigmented and depigmented nevi, the melanocytic component is usually in the form of dermal melanocytosis, including ocular melanosis, aberrant Mongolian spots, nevus of Ota, nevus spilus, and verrucose epidermal nevus [4,7,8,12-14].

Central nervous system, eye and skeletal abnormalities are the most frequent systemic complications in PPV. Our case was classified as PPV type IIa for the coexistence of port-wine stains, nevus of Ota and Mongolian spot without systemic involvement.

The pathogenesis of PPV is still unknown. Hasegawa and Yasuhara considered the entity to be a developmental abnormality of the vasomotor nerves and melanocytes derived from the neural crest [6]. Several investigators have proposed the concept of twin spotting as the genetic basis of PPV [15-17]. Happle, Steijlen and Tadini suggest that this syndrome arises as a result of a twin-spot phenomenon, namely, the occurrence of two different mutant patches involving two adjacent or corresponding areas of the body [15,18,19]. The prognosis of PPV varies between individuals [20].

The treatment of PPV is conservative [21]. PPV without systemic complications is benign and requires no treatment. The patient's quality of life may be improved by treating nevus flammeus using a pulsed dye laser and pigmentary nevi using Q-switched lasers. Some authors suggest that the pigmentary nevi should be treated first [22,23].

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