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Case Report

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Phakomatosis Pigmentovascularis: Case Report of Type IIa

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ABSTRACT

Phakomatosis Pigmentovascularis (PPV) is a rare sporadic developmental disorder characterized by coexistence of a cutaneous vascular malformation and pigmentary nevi. There are different classifications of PPV. When systemic involvement is there, a designation 'b' is used, whereas if no systemic involvement, a designation 'a' is used. Herein, we reported a 12 years old girl presented with a symptomatic persistent progressive skin lesions since birth. Systemic review and past medical history were all unremarkable. Skin examination revealed mixture of diffuse non-scaly, bleachable erythematous patches, greenish patches, and hypopigmented patches over her trunk. Ophthalmologist and neurologist consulations did not reveal any abnormalities. Based on the above clinical findings, the patient was diagnosed to have port-wine stains, Mongolian spots, and nevus anemicus. Constellation of these clinical findings without presence of extracutaneous manifestations made the diagnosis of PPV type IIa.

KEYWORDS: Phakomatosis; Pigmentovascularis.

INTRODUCTION

Phakomatosis Pigmentovascularis (PPV) is a rare sporadic developmental disorder characterized by coexistence of a cutaneus vascular malformation and pigmentary nevi. Traditionally, there are 4 types of PPV. However, later on a fifth type has been described.[1] Happle proposed a new classification of PPV composed of 4 types.[1] When there is systemic involvement, a designation 'b' is used, whereas if no systemic involvement, a designation 'a' is used. Systemic involvement is present in 50% of patients with PPV.The most common form of PPV is type II.[1-4]

CASE REPORT

A 12-year-old girl presented with a symptomatic persistent skin lesions, since birth. The lesions were increasing in size in the first few years of life but later on became stable. She did not recieve any treatment for the skin lesions. Systemic review and past medical history were all unremarkable. There was no similar case in the family and her parents are not consanguinous. Skin examination revealed mixture of non-scaly, bleachable erythematous patches, greenish patches, and hypopigmented patches over her trunk only (Figure 1). Ophthalmologist and neurologist evaluations did not reveal any abnormalities. Based on the above clinical findings, a diagnosis of port-wine stains, mongolian spots and nevus anemicus were made. Constellations of these clinical findings without presence of extracutaneous manifestations made the final diagnosis of PPV type IIa. Patient was reassured and put under periodic follow up.

DISCUSSION

The Greek word '*phakos'* means birth mark or spot. Phakomatosis is a term mainly applied to genetically determined disease characterized by the presence of oculoneurocuta-

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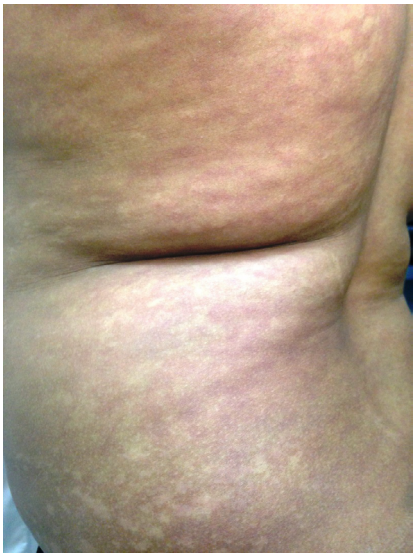

Figure 1: Mixture of diffuse non-scaly, bleachable erythematous patches, greenish patches, and hypopigmented patches over the trunk of the patient.

neous findings.[4] PPV was first described by Ota and Hasegawa in 1947. In 1985, PPV was classified into 4 types (traditional classification). Recently a fifth type has been described. Table 1 shows these 5 types of PPV. A subtype 'a' was used if there is only cutaneous involvement and subtype 'b' if there are cutaneous and extra cutaneous manifestations. Table 2 shows the systemic associations with PPV.[1]

Happle proposed a new simplified classificationof PPV.[3] Table 3 shows this new classification. In this classification, the distinction between cases that do or do not show extracutaneous anomalies is eliminated and the existence of type I. PPV is rejected on the argument that epidermal nevus never originates from pigmentary cells.[1]

The dermal melanocytosis includes Mongolian spots, nevus of Ota or nevus of Ito.[1] The pathogenesis is not completely understood. PPV may reflect twin spotting phenomenon (didymosis) as a result of hypothetic allelic mutation presented as paired melanocytic and achromic macules or nevus vascularis mixtus.[1-4]

The importance of periodic follow-up with ophthalmologist and neurologist should be emphasized, since systemic alterations can be evident with time, changing the classification and prognosis.[5,6]

The cutaneous lesions of PPV are persistent. Pulsed dye laser for nevus flammeus and Q-switched ND-Yag laser for intradermal melanocytosis have been used with good outcome.[7]

CONFLICTS OF INTEREST

The authors have no conflicts of interest that are directly relevant to the content of this case report. No sources of funding were used to assist in preparation of this manuscript.

CONSENT STATEMENT

Informed consent has been taken from the patient for purpose of using patient's photographs for publication in print or on the internet.

Type	Vascular nevus	Pigmented nevus
I	Nevus flammeus	Epidermal nevus
II	Nevus flammeus	Dermal melanocytosis ± nevus anemicus
III	Nevus flammeus	Nevus spilus ± nevus anemicus
IV	Nevus flammeus	Dermal melanocytosis + nevus spilus ± nevus anemicus
V	Cutis marmorata telangiectatica congenita	Dermal melanocytosis
Unclassified		Other associations not includedpreviously

Table 1: Classification of phakomatosis pigmentovascularis.

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Cutaneous lesions	Vascular abnormalities	Neurologic abnormalities	Ocular alterations	Miscellaneous
• Nevus anemicus • Cafe´-au-lait spots • Generalized vitiligo • Triangular congenital alopecia	• Sturge-Weber • Klippel-Trénaunay	• Seizures • Cortical atrophy • Arnold-Chiarri type I • Bilateral deafness • Idiopathic facial paralysis • Hydrocephalia • Diabetes insipidus • Plexiform neurofibroma • Delay in psychomotor development • Electroencephalogram alterations	• Melanosis oculi • Iris mammilations • Iris hamartomas • Glaucoma • Prominent vessels in sclera • Chronic edema in the cornea • Pigmentary alterations in retina • Pigmentary cataract	• Discrepancy in the length of extremities • Scoliosis • Spinal dysraphism • Hemihypertrophy • Syndactilia • Macrocephalia • Renal agenesia • Renal angiomatosis • Hepatosplenomegaly • Pyogenic granuloma • Cavernous hemangioma • Umbilical hernia • Hypoplasia of leg veins • IgA deficit • Hyper-IgE syndrome • Ezcemas • Premature eruption of the teeth

Table 2: Systemic associations with PPV.

Type	Correspondence with traditional classification	Vascular nevus	Pigmented nevus
Cesioflammea	II	Nevus flammeus	Blue spots
Spilorosea	III	Nevus flammeus	Nevus spilus
Cesiomarmorata	V	Cutis marmorata telangiectatica congenita	Blue spot
Unclassifiable	IV	Nevus flammeus	Blue spot + nevus spilus

Table 3: New classification of PPV proposed by Happle (type I does not exist).[3]

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