

# A Case Report of Phakomatosis Pigmentovascularis in a Patient with Discoid Lupus Erythematosus and Epidermal Naevus

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## Abstract

We report phakomatosis pigmentovascularis (PPV) detected in a 40- year- old male characterized by the presence of a port-wine stain in the background of aberrant Mongolian spots covering the back, nevus of Ota, ocular melanosis, epidermal nevus and a scaly patch with the diagnosis of discoid lupus erythematosus. These associations have not been reported yet.

**Keywords:** Phakomatosis pigmentovascularis, port-wine stain, dermal melanocytosis

## Introduction

Phakomatosis pigmentovascularis (PPV) refers to a rare cutaneous malformation characterized predominantly by vascular and melanocytic components<sup>1</sup>. The vascular part consists of patchy areas of port-wine stains. The melanocytic component is usually in the form of dermal melanocytosis, including ocular melanosis and aberrant Mongolian spots, verrucous pigmented nevus, or flat brown-colored plaques of nevus spilus<sup>2-4</sup>. We report a case of PPV with nevus of Ota, dermal melanocytosis, port-wine stain and epidermal nevus that can be categorized in none of the four defined types of this syndrome. In addition, according to best of knowledge, we report the first instance of PPV associated with discoid lupus erythematosus.

## Case Report

A 40-year-old white male presented with congenital vascular and pigmentary cutaneous abnormalities. On skin examination, there were three pink telangiectatic patches with irregular borders and without exophytic component over the back which were clinically compatible with port-wine stain (fig. 1). In addition, he had a blue- gray discoloration on the left side of the face associated with blue pigmentation of ipsilateral sclera. These findings are consistent with nevus of Ota (fig. 2). Similar blue-gray patches with clinical diagnosis of Mongolian spot were noted over the back (fig. 1). We found a linear brown verrucous plaque of epidermal nevus on his scrotum (fig. 3).

In addition to these congenital lesions, a well defined red scaly patch had appeared on his right



**Figure 1:** Dermal melanosis and port-wine stain over the back

supraorbital area since three months ago (fig. 2). Histopathology and direct immunofluorescence findings of this lesion revealed the diagnosis of discoid lupus erythematosus.

He had no family history of similar pigmentary or vascular defects. His parents were not consanguineous.

General physical examination including neurological and ophthalmological evaluations was unremarkable. He did not have any systemic involvements associated with lupus erythematosus or PPV. Complete blood count, liver function tests and renal function tests were normal. Anti nuclear antibodies were not found. Because of his chronic headache we performed a brain computed tomography (CT) scan which revealed no anomaly.

According to these findings, the patient had PPV and discoid lupus erythematosus simultaneously.

## Discussion

Phakomatosis pigmentovascularis refers to a rare cutaneous malformation characterized predominantly by vascular and melanocytic components. The association of cutaneous vascular malformations with pigmentary nevi was first described by Ota et al. in 1947,<sup>5</sup> who proposed the term “phakomatosis pigmentovascularis”<sup>6</sup>. The first case of PPV had a port-wine stain associated with a pigmented and verrucous nevus<sup>5</sup>. To date, only three other cases similar to this one have been published<sup>7-9</sup>. Most of the cases with PPV reported in the literature are Japanese. Hasegawa and Yasuhara<sup>4</sup> proposed a classification of PPV with

four subdivisions, each of which may be cutaneous disease only (type a) or cutaneous and systemic forms (type b). The basic alteration in each type is a port-wine stain. Nevus anemicus may or may not be present<sup>1-4,10-12</sup>. The cutaneous lesions may be the only component of the syndrome or it may be associated with systemic alterations, mainly ocular, skeletal, and neurologic<sup>2,6,10,11,13-15</sup>. The vascular portion consists of patchy areas of port-wine stain and the melanocytic component is usually in the form of dermal melanocytosis, including ocular melanosis, aberrant Mongolian spots, nevus of Ota, nevus spilus, and verrucous pigmented nevus (table 1)<sup>1,2,6,11,12,16</sup>.

In 1987, Ruiz-Maldonado et al. described four cases who were all type II; so they considered this classification too broad to be useful for the many variants which are included<sup>16</sup>. Also Happle and Steijlen considered this subdivision under dispute, because in their opinion the eight subtypes have the same genetic origin, so they cannot be separated<sup>17</sup>.

Clinical findings of nevus of Ota, ocular melanosis, port-wine stains, aberrant Mongolian spot and epidermal nevus in our patient are consistent with the diagnosis of PPV, but to our knowledge, this is a new type of this syndrome that



**Figure 2:** Left sided Ota nevus and discoid lupus erythematosus on right supra orbital area

cannot be categorized in any of previous types I-IV.

Cutaneous findings consistent with PPV require careful clinical examination, appropriate radiological investigation, and referral to other specialties, such as neurology and ophthalmology, to investigate the possibility of extracutaneous manifestations of the syndrome. No other abnormalities were observed on general physical,



**Figure 3:** Epidermal nevus on the scrotum

neurological and ophthalmological examinations in this patient, compatible with type a of PPV.

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 are described in the literature<sup>6</sup>. The real incidence of PPV is difficult to evaluate because most of the patients without visceral involvement are not reported<sup>6</sup>.

The most common systemic associations with PPV are Sturge-Weber syndrome<sup>13-15</sup> and Klippel-Trénaunay syndrome<sup>11,16</sup>. Other associated findings are temporal alopecia<sup>19</sup>, malignant colon polyposis<sup>20</sup>, scoliosis and leg-length discrepancy<sup>3</sup>, hypoplastic larynx and subglottic stenosis<sup>21</sup>, multiple granular cell tumor<sup>22</sup>, selective IgA deficiency<sup>23</sup>, iris hamartomas<sup>24</sup> and generalized vitiligo<sup>25</sup>. Although there are several associated pigmentary and systemic involvements in PPV, discoid lupus erythematosus has not been reported in association with this syndrome. These alterations are very diverse, so they may only be incidental findings and not constant associations.

**Table 1.** Classification of phakomatosis pigmentovascularis

Type	Subtypes	Characteristics
I	a or b	port-wine stain, nevus pigmentos verrucosus
II	a or b	port-wine stain, blue spots
III	a or b	port-wine stain, blue spots
IV	a or b	port-wine stain, blue spots, nevus spilus

A: cutaneous disease only; b: cutaneous and systemic disease

The pathogenesis of PPV is still controversial; it has been proposed that the combination of vascular and pigmentary anomalies arises as a result of a genetic concept called the twin-spotting phenomenon<sup>26,27</sup>.

In 1993, Happle<sup>28</sup> speculated that PPV was triggered by human mosaicism, hypothesizing that the most important factor related to the onset of PPV was an allelic mutation. The presence of perivascular nerves suggests the hypothesis that abnormal neuromodulation may play a role in the vascular component of PPV and may explain the frequent co-existence of port-wine stain and nevus anemicus<sup>28</sup>.

In conclusion, the lack of precise knowledge about the pathogenesis and the genetics of this group of diseases dose not yet allow us to develop a diagnostic algorithm and a precise scheme for the associations of the disease. However, our case is remarkable because of its unusual manifestations and its association with discoid lupus erythematosus, which has not been reported previously.

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