Phacomatosis Pigmentovascularis Revisited and Reclassified

Rudolf Happle, MD

Objective: To provide a new comprehensible and practicable classification by use of descriptive terms to distinguish the various types of phacomatosis pigmentovascularis (PPV), which has previously been classified by numbers and letters that are difficult to memorize.

Study Selection: Published case reports on PPV were reassessed.

Data Extraction and Data Synthesis: A critical review revealed that only 3 well-established types of PPV so far exist. To eliminate the cumbersome traditional classification by numbering and lettering, the following new terms are proposed: phacomatosis cesioflammea (blue spots [caesius = bluish gray] and nevus flammeus); phacomatosis spilorosea (nevus spilus coexisting with a pale-pink telangiectatic nevus), and phacomatosis cesiomarmorata (blue spots and cutis marmorata telangiectatica congenita). Phacomatosis cesioflammea is identical with the traditional types IIa and IIb; phacomatosis spilorosea corresponds to types IIIa and IIIb; and phacomatosis cesiomarmorata is a descriptive term for type V. A categorical distinction of cases with and without extracutaneous anomalies seems inappropriate. The traditional type I does not exist, and the extremely rare traditional type IV is now included in the group of unclassifiable forms.

Conclusion: The proposed new classification of PPV by using 3 descriptive terms may be easier to memorize compared with the time-honored grouping of in part not even existing subtypes by numbers and letters.

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Phacomatosis pigmentovascularis (PPV) is defined as an association of a widespread vascular nevus with an extensive pigmentary nevus. All types so far described can be best explained as examples of twin spotting. The prevailing classification of the various forms of PPV is cumbersome. How can the busy practitioner memorize the differences between type IIb and type IIIa? A recently recognized type V still adds to the confusion because there is no mnemonic value in the bare numbering and lettering of subtypes.

I propose a simplified classification by using descriptive terms. Accordingly, there are only 3 distinct types of PPV, apart from a group of unclassifiable forms. The 3 well-established forms are called phacomatosis cesioflammea, phacomatosis spilorosea, and phacomatosis cesiomarmorata (Table). The Greek word phakos means nevus. The term phacomatosis was originally used to characterize some neurocutaneous syndromes but is today mainly applied to genetically determined diseases characterized by the presence of 2 or more different nevi, such as “phacomatosis pigmentovascularis” or “phacomatosis pigmentokeratotica.” The spelling “phacomatosis” is preferable to “phakomatosis” when the term is followed by a Latin adjective.

Phacomatosis cesioflammea

This type is characterized by a coexistence of 1 or more aberrant blue spots (Mongolian spots, dermal melanocytosis) and 1 or more port-wine stains (Figure 1). The Latin word caesius means bluish gray and serves as an equivalent of the term fuscocoeroleus to describe an aberrant Mongolian spot. Phacomatosis cesioflammea is the most frequently occurring type. For example, Vidaaurri-de la Cruz and colleagues did not find any other type in a series of 24 consecutive cases of PPV.
The vascular nevus is usually purple-red and represents a nevus flammeus in the strict sense of the word. It should be noted, however, that during the first weeks of life, this port-wine stain may still show a pale-pink color.8

The 2 skin lesions may occur without other anomalies,9 or they may be associated with central nervous system defects,10 ocular anomalies such as melanosis bulbi or glaucoma,7 asymmetrical length of limbs,11,12 dysplastic veins or lymph vessels,7,15 or nevus anemicus.14,15

Table. Proposed New Classification of Phacomatosis Pigmentovascularis

<table>
<thead>
<tr>
<th>Proposed New Name</th>
<th>Type of Coexistent Nevi</th>
<th>Traditional Name</th>
<th>Reported Additional Skin Lesions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Phacomatosis cesioflammea</td>
<td>Nevus cesius (blue spot) and nevus flammeus</td>
<td>PPV type Ila/b</td>
<td>Nevus anemicus, areas of hairlessness, hypoplastic nails</td>
</tr>
<tr>
<td>Phacomatosis spilorosea</td>
<td>Nevus spilus (speckled lentiginous nevus) of the macular type and telangiectatic nevus of a pale-pink type</td>
<td>PPV type IIa/b</td>
<td>Areas of hairlessness, granular cell tumors, lymphedema</td>
</tr>
<tr>
<td>Phacomatosis cesiomarmorata</td>
<td>Nevus cesius (blue spot) and cutis marmorata telangiectatica congenita</td>
<td>PPV type Va/b</td>
<td>None</td>
</tr>
<tr>
<td>Phacomatosis pigmentovascularis, unclassifiable type</td>
<td>Various types of vascular and pigmentary nevi, sometimes with overlapping phacomatosis cesioflammea and phacomatosis spilovascularis</td>
<td>PPV type Va/b and no name</td>
<td>Café au lait macules, hypomelanotic macules, nevus anemicus, nevus sebaceous</td>
</tr>
</tbody>
</table>

Abbreviation: PPV, phacomatosis pigmentovascularis.

Figure 1. Phacomatosis cesioflammea: blue spots coexisting with port-wine stains (courtesy of Ramón Ruiz-Maldonado, MD National Institute of Pediatrics, Mexico City, Mexico).

**Figure 2.** Phacomatosis spilorosea: nevus spilus (speckled lentiginous nevus) associated with a pale-pink telangiectatic nevus (reprinted with permission from Hautarzt.2 Copyright 1989, Springer-Verlag GmbH & Co KG).

**PHACOMATOSIS SPILOROSEA**

This type of PPV is characterized by the coexistence of nevus spilus (speckled lentiginous nevus) of a macular type and a telangiectatic nevus, which usually appears, throughout life as a pale-pink color like that of a salmon patch and unlike the purple hue of nevus flammeus (port-wine stain) (Figure 2).2,16,17

Hence, the present classification implies that 2 different types of lateral telangiectatic nevi exist, which are both associated with a specific type of PPV. It should be noted that authors who have reported cases of PPV have not discriminated between these 2 types of vascular nevi but have confusingly and interchangeably used the term *nevus flammeus* in all patients.16-19 It is important to realize that the name *nevus flammeus* (port-wine stain) does not apply to the rosé-wine nevus as observed in phacomatosis spilorosea. The term *capillary malformation*20 is likewise unsuitable because it does not differentiate between nevus flammeus and other types of vascular nevi.

The term *spilus* is a short Latin word to describe speckled lentiginous nevus. The name *phacomatosis spilorosea* is proposed because *roseus* means light red or pink. The disorder may occur without other anomalies,18 or it may be associated with unilateral lymphedema,2 hypoplasia,3 hemiparesis,3 seizures,3 or asymmetrical length of legs resulting in scoliosis.17,22 Remarkably, however, neither ne-
vus flammeus nor nevus anemicus appear to be associated with this type of PPV.

**PHACOMATOSIS CESIOMARMORATA**

This type of PPV is characterized by a coexistence of nevus caesius (blue spot, aberrant Mongolian spot) and cutis marmorata telangiectatica congenita (Figure 3). The name phacomatosis cesiomarmorata contains a brief description of the 2 skin disorders. Enjolras and Mulliken described a 3-month-old boy with this new “type V” PPV. Torrelo and colleagues observed 2 additional cases, one of which was associated with blue sclerae and slightly diminished corneas. The other case involved hyperplasia of a leg, and a computed tomographic scan showed asymmetry of hemispheres and ventricles. Hence, phacomatosis cesiomarmorata may likewise be associated with extracutaneous defects.

**WHY ONLY 3 WELL-DEFINED TYPES?**

The traditional type I has not emerged in practice, and the conventional type IV also seems virtually absent. The historic type I was defined as concomitant nevus flammeus and nevus pigmentosus et verrucosus, a term that means a linear epidermal nevus with some pigmenta-

The designation “type I” has even been used to describe an unusual co-occurrence of nevus flammeus and Becker nevus. Because neither linear epidermal nevi nor Becker nevus originate from pigmentary cells, the name phacomatosis pigmentovascularis is unsuitable. In other words, type I does not exist.

Type IV was defined as an admixture of types II and III. Apparently, such overlapping phenotypes are extremely rare. It seems unnecessary to create a particular term for such cases that show a protean variability. For practical purposes it is preferable to categorize them within a group of unclassifiable PPV and to describe the particular association of anomalies as observed in a given case (Table).

**WHY NO DISTINCTION BETWEEN SUBTYPES “a” AND “b”?**

A categorical separation between cases showing exclusively cutaneous changes and those characterized by additional extracutaneous anomalies seems inappropriate. First, in other genetic disorders such as incontinentia pigmenti, cutis marmorata telangiectatica congenita or neurofibromatosis 1, we do not make this distinction either. Second, we can never be sure that a supposed type “a” will later not turn out to represent type “b.” Third,
the additional letter “b” does not contain any information regarding the question of which particular extracutaneous anomalies are present. Therefore, it seems preferable to give an exact description, if necessary, of the associated extracutaneous defects.

**CASES OF UNCLASSIFIABLE PPV**

Some cases of PPV cannot yet be ascribed to any well-defined clinicogenetic entity. Several authors reported telangiectatic nevi associated with café au lait macules (Renschler et al., 1989; Delaporte et al., 1991; Eichenfield et al., 1993; Fishman and Mulliken, 1993). Bielsa and colleagues (1993) described speckled lentiginous nevus associated with nevus anemicus. Bielsa and colleagues described 5 cutaneous components in the form of telangiectatic nevi, blue spots, café au lait macules, nevus depigmentosus, and nevus sebaceus. Bielsa and colleagues described speckled lentiginous nevus associated with nevus anemicus in a patient with bilateral primary lymphedema. From this group of unclassifiable cases, additional well-defined types of PPV may emerge in the future.

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**Correspondence:** Rudolf Happle, MD, Department of Dermatology, University of Marburg, Deutschhausstrasse 9, 35033 Marburg, Germany (happle@med.uni-marburg.de).

**REFERENCES**