Phacomatosis pigmentovascularis (PPV) represents an umbrella term for binary skin conditions characterized by the co-occurrence of a widespread telangiectatic naevus (naevus flammeus, naevus roseus (NR) or cutis marmorata telangiectatica congenita) and a pigmentary naevus (blue spot, naevus spilus maculosus or café-au-lait patches) (1, 2). The traditional classification was revised by Happle (2) and 3 major types were redefined: phacomatosis cesioflammea (3), phacomatosis spilorosea (4) and phacomatosis cesiomarmorata (5). Phacomatosis melanorosea (PMR) is a relatively novel entity belonging to the broad spectrum of PPV, anecdotally described as the coexistence of NR and one or multiple lateralized café-au-lait macules (6–10). NR, the vascular hallmark of PMR, is typically lateralized, tends to be arranged in a chequer-board pattern, and is distinguished from dark-red to purple-coloured naevus flammeus by its light-red or pale-pink colour (11). The former can also be seen as a component of phacomatosis spilorosea, which is distinct from PMR by the presence of macular naevus spilus (4).

We report here a case of an otherwise healthy 3-year-old boy with a constellation of large hypermelanotic patches and telangiectatic macules, mostly consistent with NR. Moreover, blue spots and a lesion reminiscent of segmental lentiginosis were noted. We think that this uncommon manifestation best fits into the tentative diagnosis of PMR, and briefly review the literature.

**CASE REPORT**

Our patient, now 3 years old, presented at the age of 6 months with a combination of systematized pigmentary and vascular skin lesions that had been noted at birth. He was the only child born from non-consanguineous healthy parents after an uneventful pregnancy. His medical history was unremarkable. On dermatological examination there was a large café-au-lait coloured patch with an irregular border on the right half of the trunk, largely respecting the midline. There were pale-pink telangiectatic macules forming irregular margins on the anterior and posterior aspect of the trunk. These pigmentary and vascular naeves were partly intermingled and both arranged in a chequer-board pattern (Figs 1 and 2a). In addition, there were 4 approximately 15 × 10–55 × 30-mm-sized more or less rectangular café-au-lait macules on the left arm that were slightly darker than the large pigmented patch on the trunk (Fig. 1a). He also had light-red macules distributed on the right side of his face (Fig. 2b), and macules with a more purplish hue in the nuchal area and on the right side of the neck (Fig. 1b). Two blue spots were noted; a large one located in the gluteal region and another smaller one on the dorsum of the left foot. He also had clustered punctate light-brown macules reminiscent of agminated lentigines on the left lateral aspect of the thorax (Fig. 2c). Ophthalmological and neurological work-up were normal. The patient was in good general health and exhibited normal growth and development. No change or progression was observed in the size and shape of the lesions during 2.5 years of follow-up, except for enlargement proportionate to the body size.

**DISCUSSION**

PMR was first proposed as a neologism to describe the combination of NR and large café-au-lait patches in an 11-year-old girl who also had heterochromia of scalp hair (7). A 2-month-old girl with a similar constellation of skin lesions was reported by Aguayo-Leiva et al. (8). These authors, however, preferred the term “phacomatosis melanovascularis” over PMR, arguing that NR was accompanied by cutis marmorata-like lesions in their case. In both of these cases, the pigmentary and vascular features were arranged in a chequer-board pattern, being in close apposition and partly superimposed. The third case, described as PMR, had a different clini-
Short communication

NR, the vascular component of PMR, is a laterali-
zed telangiectatic naevus with a light-red or pale-pink
colour. It differs from naevus flammeus, which also
tends to be arranged in a chequer-board pattern but
characteristically has a dark-red hue. Furthermore,
a naevus flammeus is associated with phacomatosis
cesioflammaea, whereas NR can be seen within the
context of both phacomatosis spilorosea and PMR,
supporting the view that NR and naevus flammeus
are distinct entities. However, NR and naevus flammeus
may be indistinguishable early in life, since both may
be pink-coloured during infancy, indicating the need for
long-term follow-up to make a clear-cut distinction (11).

We consider our case to be most appropriately clas-
sified as an example of PMR. The block-like appearance
and overlap of the large hypermelanotic patch and smaller
erythematous patches in the form of NR on the trunk
of our patient resemble the first 2 reports cited above
(7, 8). The vascular component of our patient, however,
deserves further mention. First, the nuchal lesion (Fig.
1b) has a slightly darker hue, reminiscent of naevus
flammeus. Secondly, the colour of the pink patches may
darken with time (11), although this is rather unlikely in a
3-year-old child. For the time being, we share the opinion
that the predominant vascular component of our patient
can, overall, be regarded as NR. Similarly, the blue spots
and segmental lentigines are less conspicuous than the
large café-au-lait patch on the trunk, which represents the
major pigmented component. Admittedly, the diversity of
the lesions hampers a simple and unambiguous classifica-
tion of our case. However, based on the major findings
the designation of PMR seems to be most appropriate.

The different types of PPV, including PMR, had
previously been regarded as examples of didymosis,
and tentatively explained at the molecular level by the
concept of non-allelic twin spotting resulting from post-
zygotic recombination (2, 7, 8). However, the theory
of twin spotting can no longer be upheld as a plausible
mechanism for PPV and a number of other binary geno-
dermatoses (6). The naevus flammeus of phacomatosis
cesioflammaea appears to originate from a heterozygous
GNAQ mutation (12), and the naevus cesius (aberrant
blue spot) is most likely caused by the same heterozygous
mutation that belongs to the group of “dominant dark
skin mutants” (13). If so, the theory of early post-zygotic
recombination can be excluded. Moreover, recent mole-
cular research revealed that both cutaneous components
of phacomatosis pigmentokeratotica, an entity charac-
terized by the coexistence of papular naevus spilus and
naevus sebaceus, originate from a common post-zygotic
mutation in a heterozygous state, thus excluding the
formerly postulated hypothesis of non-allelic didymosis
(14). Hence, phacomatosis pigmentokeratotica can be
classified as a “pseudodidymosis” (15), and the same
holds for the various types of PPV (6).

In conclusion, PMR is a binary skin disorder defined
as the coexistence of NR and lateralized café-au-lait
macules, and can be considered among the well-esta-
lished types of PPV. This rather novel entity may in
fact be more common in clinical practice, since many
cases have probably gone unrecognized so far. The
molecular basis of PMR is unknown.

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