

Cutis marmorata telangiectatica congenita and aberrant Mongolian spots: Type V phacomatosis pigmentovascularis or phacomatosis cesiomarmorata

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INTRODUCTION

The coexistence of cutis marmorata telangiectatica congenita (CMTC) with Mongolian spots has been reported as a distinct type of phacomatosis pigmentovascularis (PPV), namely PPV type V or phacomatosis cesiomarmorata.^{1,2} PPV type V is a rare congenital vascular anomaly, with only 7 previous cases mentioned in the literature. Our team conducted research on a newborn with CMTC and aberrant Mongolian spots and reviewed the literature.

CASE REPORT

A purplish reticulated vascular network, phlebotasia, and telangiectasia were observed at birth of a female of 39⁺³ gestational weeks, suggesting a diagnosis of CMTC. The lesions were located on the entire back, most of the buttocks, right half of the front of the trunk (with a clear demarcation from the unchanged skin right in the midline), right thigh, and left knee (Fig 1). The lesion was made more distinct when the baby was crying, during vigorous activity, and when cold. Additionally, the hyperpigmented patches, which were considered Mongolian spots, partly intermingled with the lesions of CMTC, were located on the right upper back, the mid-spine, and the lumbosacral region (Fig 2). Extremities were symmetrical, with no atrophy or hypertrophy of soft tissue. The infant showed normal mental and physical development. Her parents were not consanguineous. The first child of the family is a

Abbreviations used:

CMTC: Cutis marmorata telangiectatica congenita
PPV: Phacomatosis pigmentovascularis

healthy boy, who was 20 months old at the time of writing.

Regularly monitored laboratory, hematologic, biochemical analysis and chromosomal microarray detection were unremarkable. Histopathologic examination on the right side of the thigh found numerous dilated capillaries and venules in the dermis (Fig 3).

Given the constellation of clinical findings, PPV type V or phacomatosis cesiomarmorata was diagnosed. There are few cases reported in the literature of this kind of PPV.

In a follow-up examination when the infant was 2 months old, her mother reported that the child's legs trembled occasionally with no obvious cause. However, magnetic resonance imaging of the brain was unremarkable. The skin changes of CMTC had almost disappeared, while the Mongolian spots were persistent (Fig 4).

DISCUSSION

PPV is a rare congenital vascular anomaly characterized by the coexistence of vascular malformation with melanocytic lesions, including epidermal nevi and dermal melanocytosis. In 1985, Hasegawa and Yasuhara³ proposed classifying

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Fig 1. Skin changes of a mature infant age 3 days.



Fig 2. Skin changes of a mature infant age 3 days.

PPV into 4 major types according to the combination of a nevus flammeus and melanocytic lesion. Each type is further classified as cutaneous disease only (subtype a), or cutaneous with coexistent systemic disease (subtype b). The combination of CMTC and Mongolian spot, categorized as PPV type V, was added in 2003.¹

However, in 2005, Happle² found that only 3 well-established types of PPV so far exist. He considered that a categorical distinction of cases with and without extracutaneous anomalies seemed inappropriate and proposed a new classification of PPV by using 3 descriptive terms: *phacomatosis cesioflammea* (blue spots and nevus flammeus), which is identical to the traditional type II; *phacomatosis spilorozea*, which corresponds to type III (nevus spilus coexisting with a pale-pink telangiectatic nevus), and *phacomatosis cesiomarmorata* (blue spots and cutis marmorata telangiectatica congenita), which is a descriptive term for type V. The subgroup of type I was eliminated, and the extremely rare traditional type IV is included in the group of unclassifiable forms.

The lesions of CMTC, which tend to show a mosaic distribution in a patchy, quadrant, or unilateral manner with a sharp demarcation at the midline, support the hypothesis that CMTC is a disorder to which the lethal gene survives by

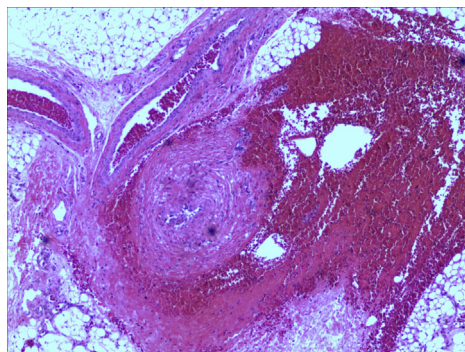


Fig 3. Histopathologic examination shows numerous dilated capillaries and venules in the dermis level. A high-resolution version of this slide for use with the Virtual Microscope is available as eSlide: VM01040.



Fig 4. Skin changes of a mature infant age 2 months.

mosaicism.⁴ The pathogenesis of PPV is still unknown. Seven cases of PPV type V (or phacomatosis cesiomarmorata) have been published, of which, 4 cases^{1,5,6} were traditional type Vb, mostly with ophthalmologic, cardiac, or brain impairment, and 3 cases⁷⁻⁹ were traditional type Va. It is interesting that the patients with type Vb all mentioned that the bluish spots and the vascular lesions faded slowly or remained unchanged. Otherwise, most of the reports of the traditional type Va did not report on follow-up. Only 1 report of a case⁹ of traditional type Va reported that both signs clearly diminished at a controlled visit after 12 months. We suspected that, in the patients with no association of systemic disease, their symptoms diminished faster. In our case, the lesions of CMTC almost disappeared in 2 months, which is faster than that of other cases, most probably because of thinning and prematuration of the lesions. It is not known if this has anything to do with the cutaneous feature in different ethnic groups, because 4 of 7 cases described in the literature are from Spain. Some investigators found differences between isolated nevi flammei and those associated with PPV, and Hagiwara et al¹⁰ report

finding endothelial cells that show neoplastic characteristics in the nevi flammei of the PPV. We suspect that there may have been differences between isolated CMTC and those associated with PPV type Va and Vb.

It is estimated that 50% of patients with PPV present with systemic involvement. Most of the studies are those associated with PPV type IIb, in which Sturge-Weber syndrome and Klippel-Trenaunay syndrome are the most common identifiable disorders, followed by central nervous system dysfunction, epilepsy, renal agenesis, and hemangioma.⁷ Most cases described in the literature are from Japan, Mexico, or Argentina.

For many patients, PPV without systematic complications is a benign condition and requires no treatment. In some persistent lesions, such as nevi flammei and mongolian spots, the cutaneous abnormality may be improved by treating with lasers.

REFERENCES

1. Torrelo A, Zambrano A, Happle R. Cutis marmorata telangiectatica congenital and extensive Mongolian spots: type 5 phacomatosis pigmentovascularis. *Br J Dermatol*. 2003;148:342-345.
2. Happle R. Phacomatosis pigmentovascularis revisited and reclassified. *Arch Dermatol*. 2005;141(3):385-388.
3. Hasegawa Y, Yasuhara M. Phacomatosis pigmentovascularis type IVa. *Arch Dermatol*. 1985;121:651-655.
4. Danarti R, Happle R, König A. Paradoxical inheritance may explain familial occurrence of Cutis marmorata telangiectatica congenita. *Dermatology*. 2001;203(3):208-211.
5. Chang BP, Hsu CH, Chen HC, Hsieh JW. An infant with extensive Mongolian spot, naevus flammeus and cutis marmorata telangiectatica congenita: a unique case of phacomatosis pigmentovascularis. *Br J Dermatol*. 2007;156(5):1068-1071.
6. Larralde M, Santos-Muñoz A, Rodríguez Cáceres M, Ciardiullo A. Phacomatosis pigmentovascularis type Va in a 3-month old. *Pediatr Dermatol*. 2008;25(2):198-200.
7. Torrelo A, Zambrano A, Happle R. Large aberrant Mongolian spots coexisting with cutis marmorata telangiectatica congenita (phacomatosis pigmentovascularis type V or phacomatosis cesiomarmorata). *J Eur Acad Dermatol Venereol*. 2006;20(3):308-310.
8. Fernández-Guarino M, Boixeda P, de Las Heras E, Aboin S, García-Millán C, Olasolo PJ. Phacomatosis pigmentovascularis: Clinical findings in 15 patients and review of the literature. *J Am Acad Dermatol*. 2008;58:88-93.
9. Cifuentes L, Kollmar A, Ring J, Mempel M. Phacomatosis cesiomarmorata in an 8-month-old infant. *Int J Dermatol*. 2009;48(10):1110-1112.
10. Hagiwara K, Uetazo H, Nonaka S. Phacomatosis pigmentovascularis type IIb associated with Sturge-Weber syndrome and pyogenic granuloma. *J Dermatol*. 1998;25:721-729.