Phacomatosis Pigmentovascularis Type Vb in a Three-Year Old Boy

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Dear Editor:

We report the case of a 3-year-old boy from Russia who presented to our department with reticulated erythematous brown patches involving the face and extremities. All patches were present at birth. The mother's pregnancy and delivery were uneventful. He had no remarkable familial history. Physical examination showed erythematous mottled marble-like lesions on the face and upper extremities. Brown reticulated patches were observed mainly on the lower extremities. In addition, poorly defined gray-bluish pigmented patches suggestive of Mongolian nevi were found on the trunk. Hypertrophy of the right side of the face and leg was observed. The right leg was 2 cm longer than the left (Fig. 1). He showed normal mental and physical development. All laboratory parameters were within normal limits. Brain magnetic resonance imaging revealed an old lacunar infarct in the right centrum semiovale. Multiple skin biopsies the erythematous patch of the face and a brown patch of the leg were taken. The histopathologic findings from the erythematous patch were generally normal, except for increased melanin pigment in the basal layer and dermal capillary dilatation (Fig. 2A). The brown patch exhibited increased melanin pigment in the basal layer and spindle-shaped melanocytes with abundant melanin in the dermis (Fig. 2B). Phacomatosis pigmentovascularis (PPV)

is a rare genodermatosis characterized by the coexistence of pigmentary abnormalities and cutaneous vascular malformation. The "twin-spot phenomenon" is a possible cause of PPV, but the exact genetic and molecular bases of PPV are unclear. PPV is classified into five types according to distinguishing features; moreover, all types are subdivided into subtype "a" and "b", indicating PPV with oculocutaneous involvement only and extracutaneous involvement (e.g., neurologic and skeletal symptoms), respectively^{1,2}. Type V PPV, described by Torrelo et al.¹ in 2003, is characterized by cutis marmorata telangiectatica congenita (CMTC) associated with aberrant Mongolian nevi. The diagnosis of PPV is made clinically. To our knowledge, only seven patients with PPV type V have been reported in the literature^{3,4}, none of which were in the Korean literature. Among patients with PPV type V, the most frequently associated extracutaneous features were ophthalmic abnormalities including ocular melanosis, glaucoma, leukocoria, and corneal opacity^{1,2,4}. Body asymmetry and asymmetry of the brain hemispheres have also reported^{1,2}. Although there were cases of cerebral ischemia patient associated with CMTC, there was no definite evidence of an association between cerebral ischemia and CMTC⁵. As observed in our patient, PPV is a congenital abnormality that can be associated with not only ophthalmic abnormality but also skeletal and neurological abnormalities. Considering extracutaneous abnormalities, it is important patients receive a full physical examination and proper management immediately after PPV diagnosis and undergo regular follow-up. Although CMTC and Mongolian spot, both tend to lighten over time, prominent lesions have remained in many cases¹⁻⁴. Pulsed dye laser and Q-switched lasers for CMTC and pigmented nevi, respectively, can be helpful for improving aesthetics. In summary, we report a rare case of a 3-year-old boy diagnosed with PPV type Vb.

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Fig. 1. (A~C) Reticulated, mottled, marble-like erythema on the face and arms, reticulated brown patches on the legs. In addition, bluish-grey patches are seen on the trunk and thighs. (D) The full leg X-ray shows discrepancy in length between the right and left leg.

Fig. 2. (A) Increased melanin pigment in the basal layer and dilation of capillary in the dermis (H&E, ×100). (B) Increased melanin pigment in the basal layer and scattered spindle-shaped melanocytes with melanin pigment in the dermis (H&E, ×100).

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