

CASE REPORT

Unilateral Nevoid Telangiectasia Associated with Ipsilateral Melorheostosis

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Melorheostosis is a rare disorder characterized by irregular, flowing hyperostosis in long bones, commonly described on radiographs as wax flowing down a candle. In addition to bony sclerosis, cutaneous manifestations overlying the involved bones have been reported including linear scleroderma, neurofibromatosis, and vascular and lymphatic malformations. Unilateral nevoid telangiectasia (UNT) is a rare primarily cutaneous condition characterized by linearly arranged small dilated blood vessels in dermatomal or Blaschkoid patterns on the skin. Here, we present the case of a nine-year-old Korean male with UNT associated with ipsilateral melorheostosis. (**Ann Dermatol 24(2) 206 ~ 208, 2012**)

-Keywords-

Melorheostosis, Nevoid, Telangiectasia, Unilateral

INTRODUCTION

Melorheostosis is a rare bone dysplasia characterized by localized hyperostosis and sclerosis, which usually affects unilateral long tubular bones¹. Reported cutaneous manifestations overlying the involved bones in patients with

melorheostosis include linear scleroderma, neurofibromatosis, and vascular and lymphatic malformations². Unilateral nevoid telangiectasia (UNT) is a rare primarily cutaneous condition characterized by linearly arranged small dilated blood vessels in dermatomal or Blaschkoid patterns on the skin³. Here, we describe the case of a nine-year-old Korean male, who presented with both melorheostosis and UNT.

CASE REPORT

A nine-year-old male patient visited our clinic with multiple asymptomatic red to bluish macules on the left chest wall and left arm, which had persisted since birth. The boy's medical history as well as that of his family was unremarkable with no reports of hepatic disease or endocrinopathy. A physical examination revealed multiple dermatomal arranged linear telangiectasias on his left chest wall and left arm (Fig. 1). In addition, a hard bony mass was noted on his left wrist and his left arm length was shorter than that of his right arm. Roentgenographic examination of the left arm was performed and showed diffuse bony sclerotic changes in the distal humerus, ulna, carpal, and second to fourth metacarpal and phalangeal bones (Fig. 2A, B). Bone scintigraphy showed abnormal uptake in the left humerus, radius, and hand bones (Fig. 2C). Based on the roentgenographic and bone scintigraphy findings, the patient was diagnosed with melorheostosis.

A skin specimen obtained from the lesions on the left arm showed numerous dilated thin-walled, small-sized blood vessels in the papillary dermis (Fig. 3). Endothelial cells of the dilated blood vessels presented neither signs of vascular proliferation, nor pathologic findings of inflammatory cell infiltration. Based on the clinical features and histopathologic and radiographic findings, a diagnosis of UNT associated with ipsilateral melorheostosis was made.

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Fig. 1. (A) A nine-year old boy with dermatomal arranged linear telangiectasias on the left chest wall and left arm. (B) Magnification of the chest lesion shows linearly arranged erythematous telangiectasias (white arrowheads) on the left chest.



Fig. 2. (A, B) Roentgenographic examination of the left arm shows diffuse bony sclerotic changes on the distal humerus, ulna, carpal, and second to fourth metacarpal and phalangeal bones. (C) Bone scintigraphy shows abnormal uptake in the left humerus, radius, and hand bones.

DISCUSSION

Melorheostosis was first described by Leri and Joanny¹ in 1922 and more than 250 cases have been reported since. In a review of 131 cases⁴, associated skin manifestations of melorheostosis were reported in 22 cases (16.8%) and

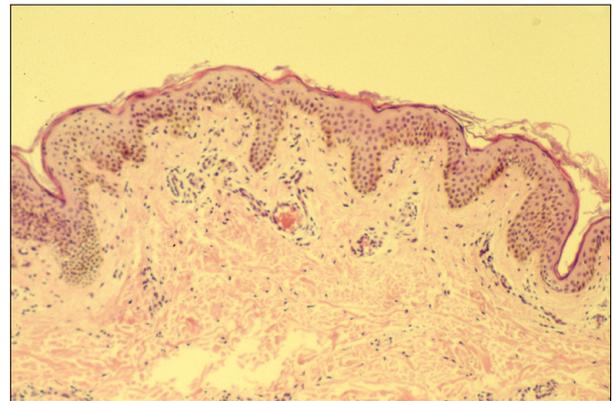


Fig. 3. Histopathology shows numerous dilated thin-walled, small-sized blood vessels without endothelial proliferation and vasculitis in the papillary dermis (H&E, ×100).

described as scleroderma, neurofibromatosis, lymphatic and vascular lesions, and skin pigmentation. Cutaneous vascular lesions, including hemangioma, enlarged surface capillaries, arteriovenous aneurysm, vascular nevus, and glomus tumor, occurred in seven instances (5%).

Although the precise pathogenesis remains to be elucidated, there have been several proposed causes of melorheostosis. Primary developmental defects in mesodermal cells have been suggested as a major pathogenic factor in melorheostosis⁵. Murray and McCredie⁶ proposed that bony lesions are a late consequence of a segmental sensory nerve lesions that account for the sclerotomal distribution of the hyperostosis associated with the disorder. Currently, postzygotic mutation during embryogenesis is thought to contribute to the congenital nature and linear distribution of the lesions⁷.

UNT is characterized by unilateral dermatomal distri-

bution of primary telangiectasia, which may be congenital or acquired. While the exact pathogenesis is unclear, an increased level of estrogen or estrogen receptors is accepted as the most possible cause of the acquired form⁸. However, there have been cases without significant elevation of estrogen levels and abnormal liver function tests. Therefore, considering that most nevoid skin diseases can be explained by genetic mosaicism, somatic mosaicism may be a more plausible cause than hyperestrogenemia in such cases.

Twin spotting refers to two recessive mutations on either pair of homologous chromosomes, which may lead to formation of two different homozygous daughter cells with two different mutant lesions by mitotic cross-over⁹. Because both melorheostosis and UNT can have mosaicism as a common pathogenic origin, the coexistence of these two rare entities can also be explained by twin spotting, which was proposed by Happle⁹ as a potential mechanism for the development of coexisting birthmarks. In addition, previous reports have described cases of twin spotting associated with either melorheostosis or UNT^{7,8}. In this report, we present a patient with both melorheostosis and UNT. We also suggested that the association between the two conditions may be explained by twin spotting.

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