Maffucci's Syndrome

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A case of Maffucci's syndrome is reported in a 6-year-old girl. She had multiple enchondromas of the long bones and the phalangeal bones with hemangiomatous skin lesions. The hemangiomas were the capillary type and were not anatomically related to the bone lesions. (Ann Dermatol 4 : (1) 37−40, 1992)

Key Words : Maffucci's syndrome, Enchondroma, Capillary hemangioma

Maffucci's syndrome, or dyschondroplasia with hemangiomata, is a rare disorder with no recognized genetic basis, characterized by general mesenchymal neoplasia that has a propensity for malignant transformation.[1] The incidence of this syndrome is relatively rare; a few cases have been reported in the Korean literature.[2−5] We present herein a case of Maffucci's syndrome with the typical bone lesions but less prominent vascular tumors.

REPORT OF A CASE

A 6-year-old girl has had multiple bony protrusions and vascular skin lesions since early infancy. At birth, she had no apparent anomalies, but a swollen right foot was noted 3 months later. At 1 year of age, multiple bony masses had developed on the digits of the hands and feet and the long bones, and a few vascular skin lesions had appeared on her flank, inguinal area, labium major and the back(Fig. 1, 4).

On physical examination she had normal facies and stature but unequal length of her extremities. There were no abnormalities in either her neurologic examination or laboratory investigations. The bone lesions were symmetric to some degree and radiologic findings showed radiolucent densities and multiple metaphyseal irregularites(Fig. 2). Bone biopsy of the distal end of the right tibia was diagnosed as an enchondroma(Fig. 3). There were five skin lesions, all small(less than 0.8㎝ in diameter), smoothly surfaced reddish nodules. The specimen obtained from the labium lesion showed numerous irregularly shaped vascular lumina and considerable proliferation of the endothelial cells(Fig. 5). The other specimen from the flank showed slight epidermal acanthosis and greatly dilated vascular cavities lined with single layer of flattened endothelial cells near the epidermis(Fig. 6).

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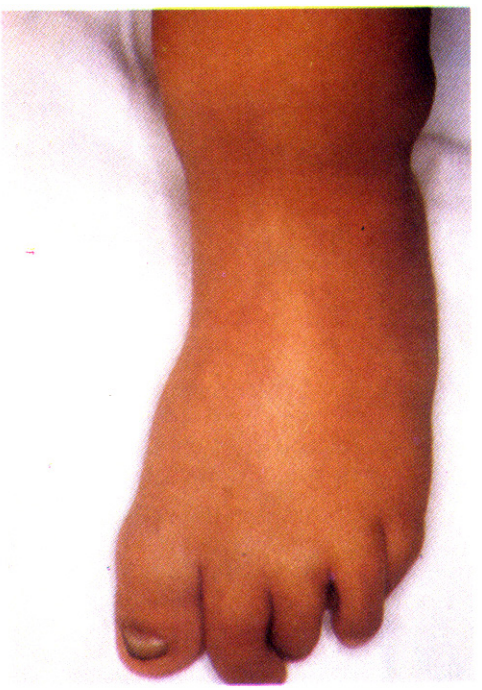

Fig. 1. Anomalous left foot with multiple bony protrusions.

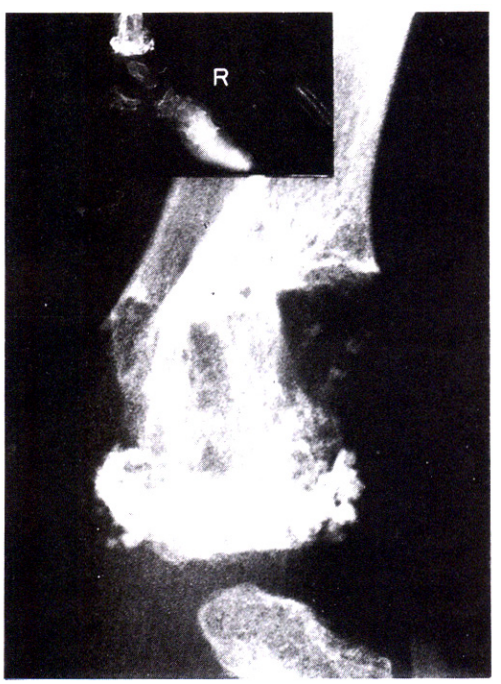

Fig. 2. Multiple metaphyseal irregularities with calcified spots in the metaphyses and epiphyseal plates.

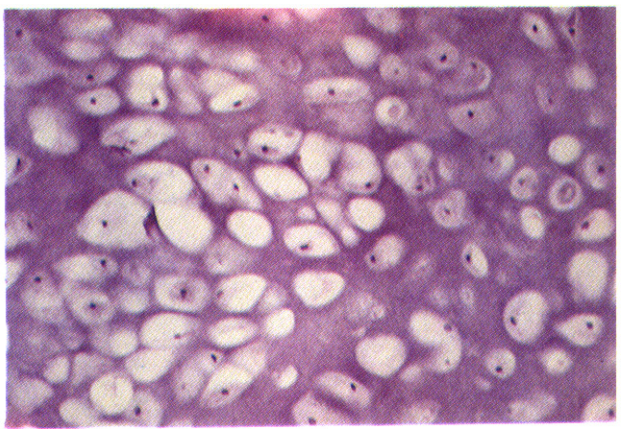

Fig. 3. Bone biopsy showing cartilage cells(H & E stain, ×200).

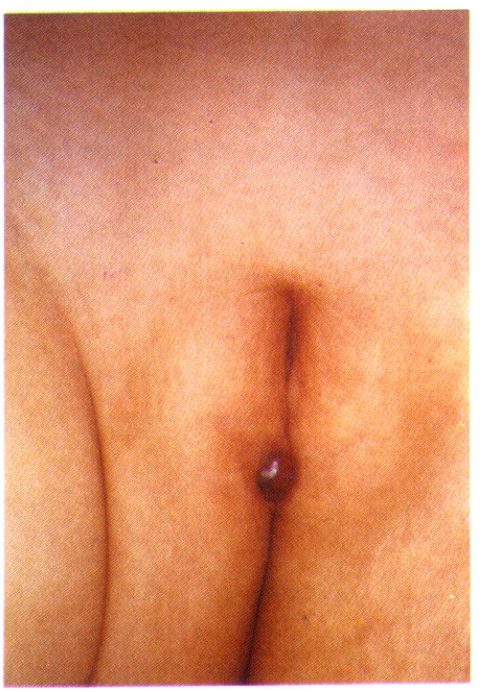

Fig. 4. 0.8cm×0.8cm sized smoothly elevated vascular nodule in the right labium major.

DISCUSSION

This uncommon syndrome was delineated by Maffucci in 1881 as multiple angiomas and enchondromas. Enchondromatosis or dyschondroplasia results from cartilage failing to undergo the normal process of endochondrial bone

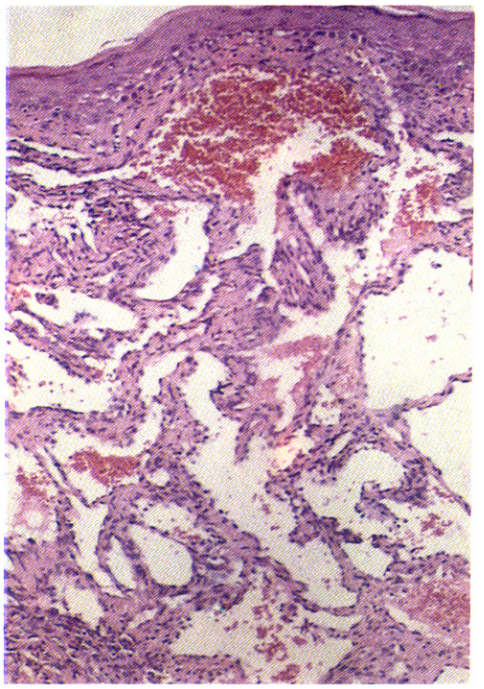

Fig. 5. Biopsy specimen from the labium showing numerous irregular vascular lumina and endothelial cell proliferation(H & E stain, ×100).

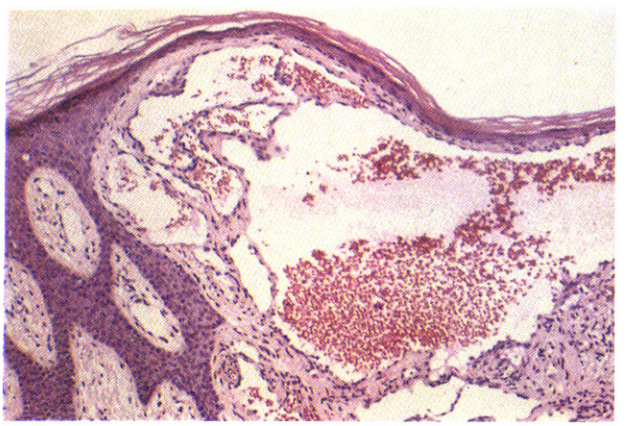

Fig. 6. Biopsy specimen from the flank showing epidermal acanthosis and a greatly dilated vascular cavity near the epidermis(H & E stain, ×100).

formation. Round masses or columns of uncalcified cartilage are produced within the metaphyses and diaphyses of certain bones, which are invariably shortened.[6] The dyschondroplasia usually appears later than the vascular lesions. Dyschondrotic changes and cutaneous hemangiomas exist independently and do not necessarily occur in the same location.[7,8] This corresponds to our case, in which hemangiomas occurred primarily on the trunk. The vascular lesions include hemangiomas, phlebectasia, vascular hamartomas and lymphangiomas. Among these, cavernous and capillary hemangiomas are the most common cutaneous findings.[1] Other cutaneous features are pigmentary changes such as vitiligo, cafe-au-lait macules, hyperpigmentation, and nevi.[9]

In our patient, one skin biopsy specimen from the labium showed a capillary type hemangioma. Although there were cavernous spaces in the deep portion of this tumor, it could be diagnosed as a capillary hemangioma because of their existence in the usual capillary hemangioma of infancy in addition to the lobulated pattern and the endothelial cell proliferation of the lesion.[8] The other specimen from the flank revealed the epidermal changes, i.e. acanthosis, which suggested angiokeratoma circumscriptum. However, it is difficult to regard the above finding as true angiokeratoma lesion because the acanthotic change was so mild and there were no other epidermal changes such as hyperkeratosis and papillomatosis. Furthermore, the appearance of the lesion was far from that of an angiokeratoma.

The skin and skeletal anomalies of Maffucci's syndrome are usually bilateral but asymmetrical and they develop during the first decade or so of life. By the second decade, the disorder usually remains stable.[10]

The diagnosis of Maffucci's syndrome is usually self-evident. Brain scan and computed axial tomography may be necessary to identify intracranial involvement.[1,9] The most serious complications occuring in this syndrome, aside from the multiple fractures in childhood, are multiple neurologic deficits resulting from cerebral encroachment of enchondromas of the skull and the development of malignant tumors. Chondrosarcoma is the most common malig-

nant tumor that occurs, in about 20% of the patients.[1]

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