

CASE

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Multiple Nodules and Masses on the Hand of a Girl

A 12-year-old girl was visited at our clinic with multiple soft tissue and bony painless swellings over her right hand since she was 2 years old (Figure 1). Soft tissue masses increased in size and number. She was born to non-consanguineous parents. Her early development was normal and she had no other medical problems. There was no family history of similar problem.

Examination revealed multiple compressible soft tissue masses and hard nodules of varying size over fingers and manus of the right hand with superficial hyperkeratosis. There was no limb asymmetry. Her height and weight were within normal range. Examination of parents was normal.

X-ray of the right hand is shown in figure 2. A biopsy specimen was taken from a skin nodule (figure 3). (*Iran J Dermatol 2010;13: 31-32*)

What is your diagnosis?

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Figure 1. Multiple soft tissue masses and hard nodules on fingers and manus of the right hand with superficial hyperkeratosis.



Figure 2. Right hand X-ray; osteolytic lesions in the 5th metacarpus and proximal phalanges of the 4th and 5th finger, compatible with enchondromas, and soft tissue masses.

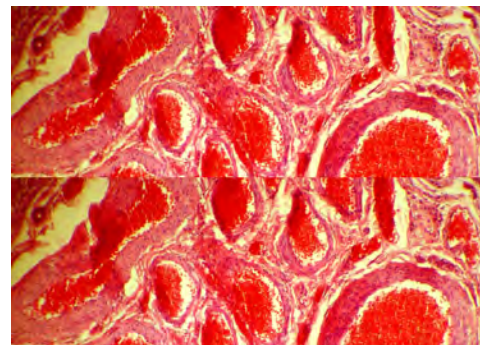


Figure 3. Dilated and congested vascular spaces, compatible with hemangioma (H&E*10).

Diagnosis: Maffucci's syndrome

Right hand X-ray revealed multiple lytic lesions in the 5th metacarpus and proximal phalanges of the 4th and the 5th finger, compatible with enchondromas, as well as several soft tissue masses (Figure 2). Histopathologic examination of one of soft tissue masses demonstrated dilated and congested vascular spaces, compatible with hemangioma (Figure 3).

According to multiple hemangiomas in association with multiple enchondromas, the diagnosis of Maffucci's syndrome was made. The patient has been carefully followed up since then.

Discussion

Maffucci's syndrome is a rare congenital non-hereditary mesenchymal dysplasia¹ which was first described in 1881. Since then, about 200 cases have been described in the literature. This syndrome is characterized by multiple enchondromas, hemangiomas and less often, lymphangiomas^{2,3} occurring during the first two decades of life⁴.

Patients are usually asymptomatic at birth. The average age at which lesions appear is 5 years, but there are reported cases stating that symptoms can occur in the first year of life¹. No familial pattern and racial or sexual predilection has been detected in this syndrome.

Enchondromas are expanding cartilaginous tumors occurring in the metaphysis of tubular bones primarily in hands, feet and long bones but may also involve the humerus, tibia, fibula, ribs or cranium⁵.

Soft tissue hemangiomas in Maffucci's syndrome are mostly located in the subcutaneous tissues and manifest as deep blue nodules which can be drained by direct pressure³. Both cavernous and capillary hemangiomas have been seen in this syndrome but the cavernous type is more common⁴.

The risk of malignant transformation is about 25%. Sarcomatous transformation may be seen in bones and soft tissue lesions but is more frequently

found in enchondromas with an incidence ranging from 15 to 57%^{2,6}.

Pathological fractures are found in 26% of the cases; other possible complications are hemorrhage, short stature, pleural effusion² and cranial nerve paralysis due to endochondral compression⁷.

The only differential diagnosis of Maffucci's syndrome is Ollier's disease which is characterized by isolated enchondromas without hemangiomas. The differentiation of these two conditions is essential, because although chondrodysplasia is found in both, it increases the risk of malignancy in Maffucci's syndrome³.

Treatment is usually symptomatic. Surgical procedures consist of osteotomy and curettage of the bone lesions. Other therapeutic options are sclerotherapy, irradiation and vascular lesion surgery. Excision is the procedure of choice for soft tissue tumors³.

Bone and soft tissue lesions that grow or become painful without a history of trauma should be examined for malignancy and biopsied⁶.

References

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