CASE 1

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Case

A 10-year-old girl presented with a 1-year history of an eruption on her hands. Physical examination revealed soft, bluish slightly tender subcutaneous nodules and swellings in the ulnar site and palmar aspect of five fingers of the right hand and dorsal aspect of three fingers of the left hand without any gross deformity in fingers and long bones (figure 1). History of pathological fractures was negative. X ray of the right hand showed reduction in bone density with lytic lesions in five metacarps and proximal phalanx of the fourth and fifth fingers associated with soft tissue swellings (figure 2). A biopsy specimen was taken from a skin nodule (figure 3).

What is your diagnosis?

Move on to next page for the answer and discussion.

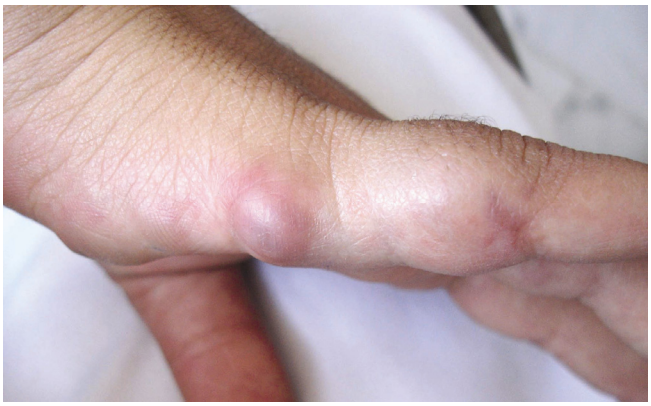

Figure 1

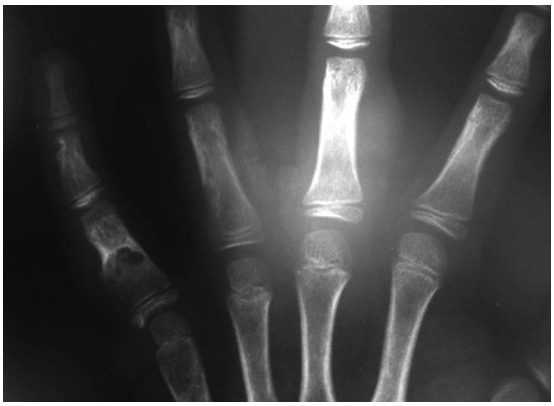

Figure 2

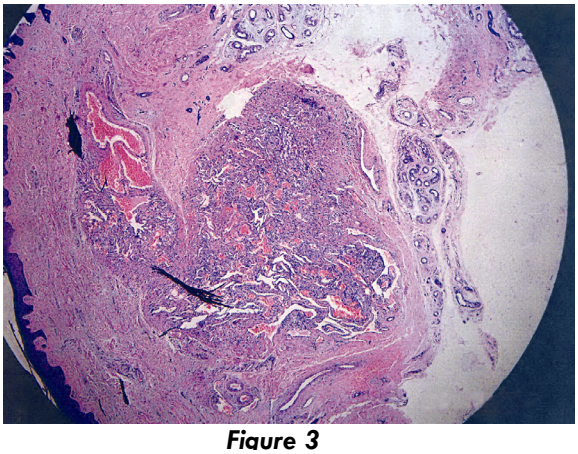

Figure 3

Diagnosis: Maffucci's Syndrome

Microscopic and imaging findings

Histologic examination revealed vascular proliferation with central branching vesseles in a lobular pattern with fibroconnective septa covered by hyperplastic epithelium consistent with haemangioma (figure 3). In MRI from the right hand, there were small soft tissue nodules with various sizes in the ulnar site and palmar aspect of the third and fifth fingers suggestive of multiple small cavernous haemangioma. Also, intra medullary lesions were seen in the proximal phalanx of the fifth finger consistent with encondroma. Similar lesions were seen in the middle and proximal phalanges of the forth finger. Overall, multiple caverneous haemangioma associated with encondromas were compatible with Maffucci's syndrome. The patient was treated with intralesional steroid and cryotherapy for cutaneous lesions and received a careful fallow-up.

Discussion

Maffucci syndrome comprises the association between cutaneous venus malformation and dyschondroplasia[1]. Such lesions were first described by Angelo Maffuci in 1881. It is a rare genetic sporadic disease[2]. The disease is the result of mesodermal dysplasia early in life without a recognized inheritance mode and genetic locus. It is seen in all races and both genders. Affected individuals are generally of normal appearance at birth, but multiple cutaneous vascular swellings start to appear in infancy which take the form of soft, bluish occasionally tender and asymmetrical subcutaneous protrusions on the hands and feet[3] and grow proportionately with the child. Histologically, skin lesions are made of subcutaneous, vascular spaces with a single endothelial lining. They do not appear to be true haemangioma but complex venous malformations[4].

Other reported cutaneous findings include cavernous lymphangioma and pigmentary changes particularly café-au-lait macules[5]. Simultaneous with the appearance of the vascular swellings, the patient develops hard nodules arising from bones, especially those of the fingers and toes, and the metaphyses of the long bones of the arms and legs. pathologically, these are enchondromas, which are radiologically translucent.

These bony lesions may be unilateral or asymmetrical. The growth of affected bones is delayed and distorted and pathological fractures and deformities are a common occurrence[4]. A variety of other benign and malignant mesodermal tumors have been reported in Maffucci syn.

Malignant transformation of enchondromas in to chondrosarcoma occurs in some 30-40% of patients[3]. Other malignant mesodermal tumors include fibrosarcoma, angiosarcoma, lymphangiosarcoma and osteosarcoma.

Ovarian tumors and non mesodermal tumors have been described including glioma and adenocarcinoma of the pancreas[6].

There are several reports on its association with breast, ovarian, pancreas, parathyroid and pituitary tumors.

Patients suffering from Maffucci syndrome without malignant transformation have a normal longevity[3]. Simple radiographic films are useful in diagnosis. Clearly these patients require a careful follow-up with a low threshold for radiological or histological assessment of any lesion that enlarges rapidly or causes symptoms. Symptomless vascular lesions are treated conservatively.

References

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