Images in Clinical Practice

Maffucci syndrome

An 18-year-old girl born of a non-consanguineous marriage presented with multiple soft tissue and bony painless swellings over fingers, toes and ankles since the age of 3½ years. Soft tissue swellings were increasing in number, size and progressively involving various sites. There was no family history of similar problem. Her early development was normal and she had average scholastic performance. Examination showed multiple hemangiomas of varying size over right side of chin, right side lower gum, right sided iliac crest, fingers, both ankles and toes, and on the plantar surface. There was no limb length asymmetry or widening of lower ends of bones. She had deformed middle finger of the right hand and ring and little fingers of left hand and right great toe due to bony swelling (*Fig. 1* and *Fig. 2*)]. Examination of parents was normal. Skeletal survey showed multiple enchondromas on phalanges and metacarpals, lower end of ulna (*Fig. 3*) and upper end of tibia.

Maffuci syndrome is a rare sporadic dysplasia

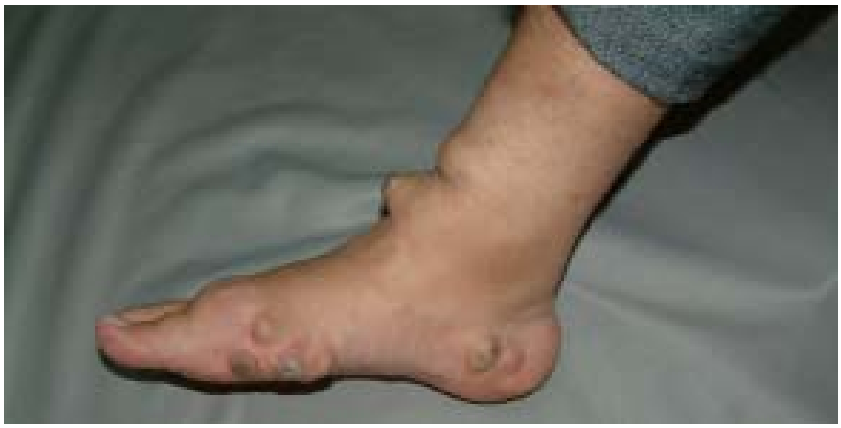

Fig. 1. Multiple hemangiomas on foot and toes.

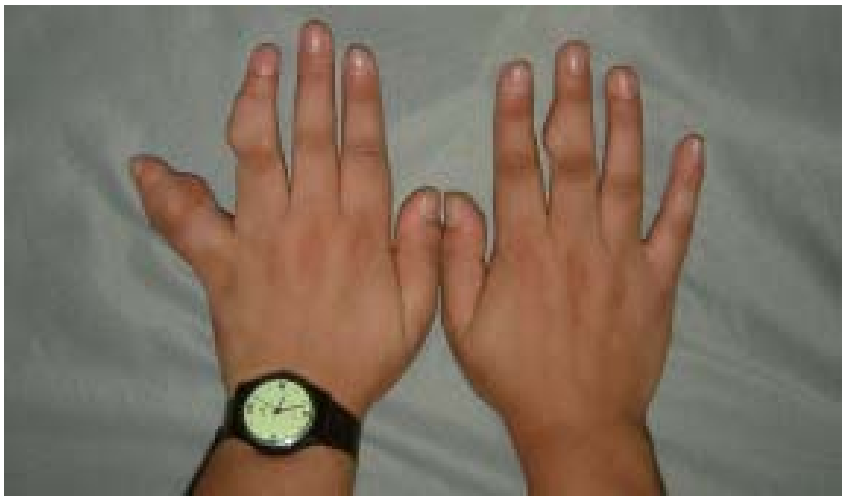

Fig. 2. Deformed fingers due to enchondromas

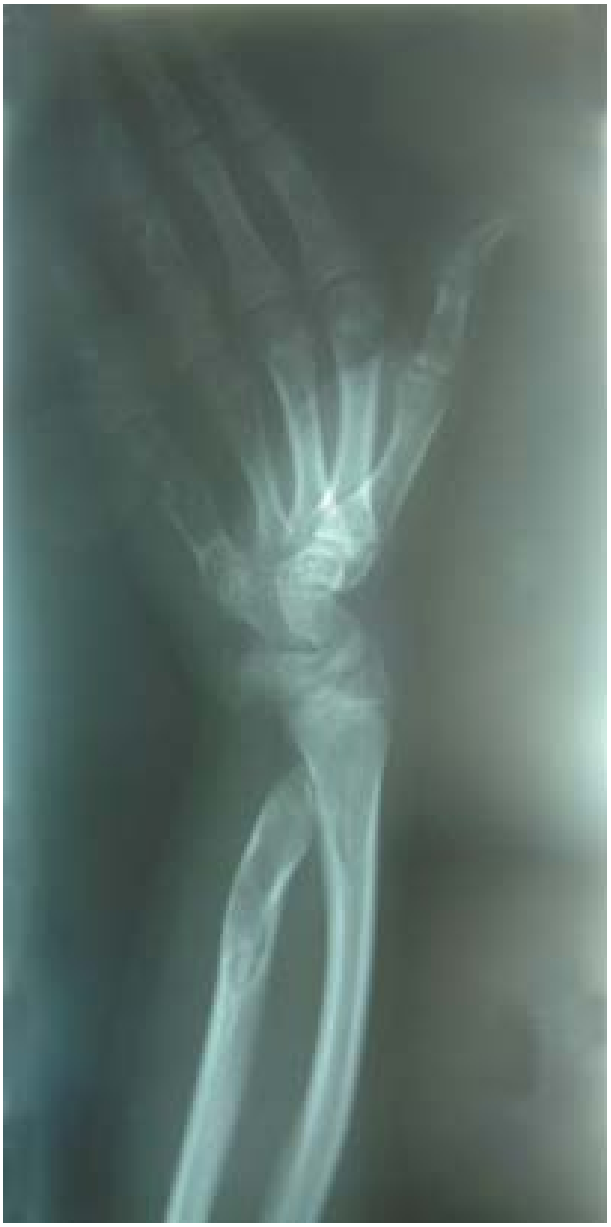

Fig. 3. Benign cartilage tumors at lower end of ulna.

characterized by superficial hemangiomas within first 4 years of life. Expanding cartilaginous tumors occur in metaphysis of tubular bones (enchondroma) primarily in hands, feet and long bones by adolescence without progression after completion of puberty. The tumors are asymmetrical and bilateral. Extra Skeletal manifestation includes presence of varying sized simple or cavernous cutaneous hemangiomas, lymphangioma, and phlebectasia. Hemangiomas are most frequently located in the dermis and subcutaneous fat adjacent to areas of enchondromatosis or else where and may show a similar distribution with regard to laterality. Complications include spontaneous fracture through area of advanced rarefaction (26%) and sarcomatous degeneration of enchondroma in 15-30%. Benign and malignant degeneration of hemangiomas and lymphangioma can also occur.

Treatment consists of orthopedic and surgical intervention to minimize deformities and for cosmetic purpose. Careful surveillance for malignant degeneration of both skeletal and non-skeletal tumors especially in the brain and abdomen is essential.

Only differential diagnosis is Ollier's disease having isolated enchondromas without hemangiomas.

Etiology of Maffuci syndrome is unknown however, mutation in the PTH/PTHRP type I receptor has been suggested. An extensive recent study showed neither abnormality of expression of PTHR1 protein by immunochemistry, nor any causative mutation in the PTHR1 gene.

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