Maffucci Syndrome: A Case Report

Raymond T. Kuwahara, MD; Robert B. Skinner, Jr, MD

The multiple enchondromas and hemangiomas of Maffucci syndrome affect the skin and skeletal systems. The disease develops slowly, with enlargement of enchondromas and hemangiomas occurring during the first 2 decades of life. Effects of the disease range from those requiring minor adjustments in activities of daily living to almost total incapacitation.

Case Report

A 35-year-old African American man presented for diagnosis of soft asymptomatic nodules on his upper extremities. The lesions had begun when he was 8 years old and had grown and spread bilaterally from his hands to upper arms. He also had firm subcutaneous nodules on his phalanges. The lesions never bothered him, and he and his physician felt that they were benign and did not require diagnosis, but the patient's fiancée asked that he have the lesions examined by a specialist.

Comment

Maffucci syndrome consists of multiple enchondromas and hemangiomas.[1] This rare syndrome was first reported by Maffucci[2] in 1881, after a 40-year-old patient had frequent and severe bleeding that led to amputation of a distal extremity. The patient died of complications secondary to infection. Maffucci described a thorough autopsy and reported all the main points of the syndrome that was to be named after him. Carleton et al[3] proposed the eponym *Maffucci syndrome* in 1942.

There seems to be no sexual or racial predilection for this syndrome.[4] No familial pattern of inheritance has been demonstrated, but the disease usually appears around the age of 5 years. Twenty-five percent of cases are congenital, and 78% begin before puberty.[4] Patients are of average intelligence, and no mental or psychiatric abnormalities have been noted. The disease seems to develop from mesodermal dysplasia early in life.

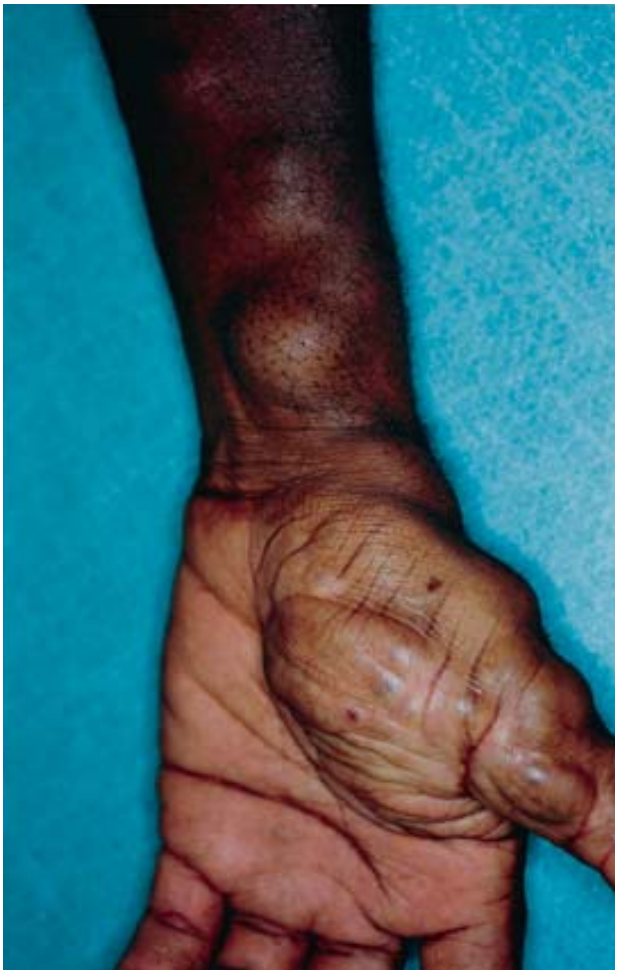

Figure 1. Large hemangiomas of Maffucci syndrome on the distal extremities.

Maffucci syndrome affects the skin and skeletal systems with multiple enchondromas and hemangiomas (Figure 1). The disease develops slowly, with enlargement of enchondromas and hemangiomas occurring during the first 2 decades of life.[5,6] Progression of these lesions usually ceases by 30 years of age.

Dr. Kuwahara is from the Department of Dermatology, University of Oklahoma, Oklahoma City. Dr. Skinner is from the Division of Dermatology, University of Tennessee, Memphis.
Reprints: Raymond T. Kuwahara, MD, Department of Dermatology, University of Oklahoma, 619 NE 13, Oklahoma City, OK 73104 (e-mail: r.kuwahara@excite.com).

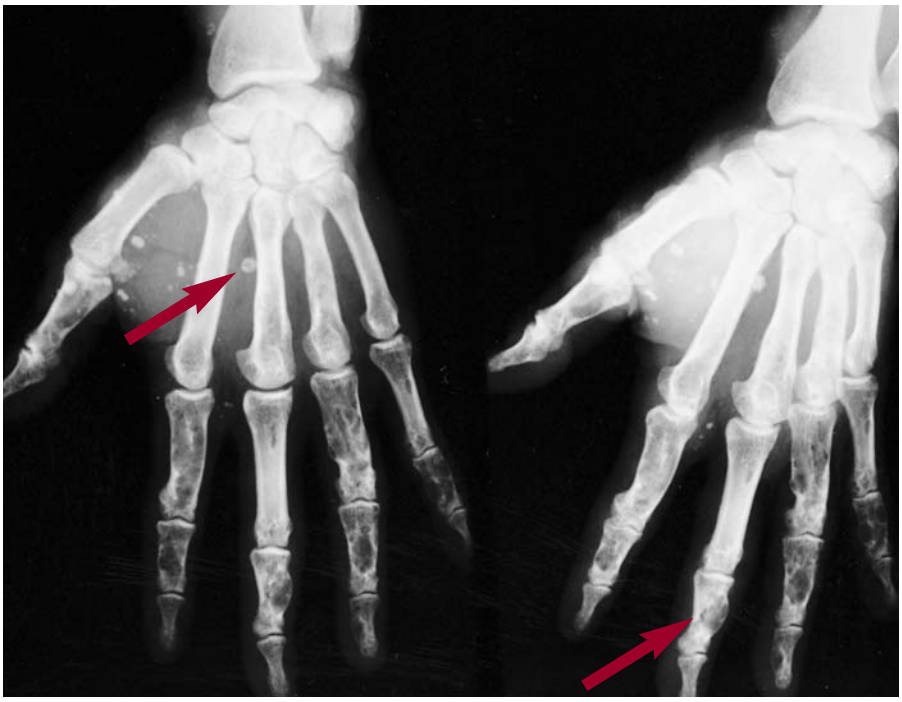

Figure 2. Our patient had multiple soft nodules on bilateral upper extremities and mildly deformed fingers since the age of 8 years.

Enchondromas are benign cartilaginous tumors that can appear anywhere but are usually found on the distal phalanges and long bones. As the bones grow, some cartilaginous material is left behind. This material grows irregularly and develops into enchondromas. These bony abnormalities are usually asymmetric and can cause secondary fractures. Enchondromas are usually found in the hands but can be found in the feet, tibias, fibulas, femurs, and skull. Patients who are severely affected may have trouble ambulating because of pathologic fractures that result in uneven limb length. Thirty percent to 37% of enchondromas associated with Maffucci syndrome develop into chondrosarcomas.[5,7] On radiographs, enchondromas appear as bony translucencies (Figure 2).

Superficial and deep venous malformations often protrude as soft nodules or tumors. Most often, they occur on distal extremities, but they can appear anywhere.[5] The hemangiomas appear clinically as bluish subcutaneous nodules that can be emptied by applying light pressure. Often, thrombi form in the lumen of the vessels—creating phleboliths. These phleboliths appear as small opacities on radiographs (Figure 2) and as calcified vessels under the microscope.

No treatment is required in mild forms of Maffucci syndrome. More severe cases involve bone deformities that make engaging in activities of daily living difficult. Wearing special shoes or using crutches may be necessary in patients who are moderately affected; patients who are severely affected may require functional amputation at the transfemoral or transhumeral level and, subsequently, prosthetic devices.

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