



## Maffucci Syndrome

Enchondroma with multiple angiomas (Maffucci syndrome) was first reported by Maffucci in 1881 after a 40-year-old woman died from complications following amputation of an arm. The patient had frequent and severe hemorrhage from a vascular tumor for which she was admitted to the hospital. In view of the profuse bleeding, an amputation was performed and the patient died from infection. Maffucci reported a thorough autopsy that described all the main points of the syndrome named after him. In 1941, Carleton et al proposed the eponym Maffucci syndrome.

Maffucci syndrome is a rare genetic disorder that affects both males and females. Maffucci syndrome is characterized by benign enlargements of cartilage (enchondromas); bone deformities; and dark, irregularly shaped hemangiomas. No racial or sexual predilection is apparent in Maffucci syndrome. No familial pattern of inheritance has been shown, but Maffucci syndrome manifests early in life, usually around age 4-5 years, with 25% of cases being congenital. Maffucci syndrome appears to develop from mesodermal dysplasia early in life. Patients apparently are of average intelligence, and no associated mental or psychiatric abnormalities seem to be present. About 160 cases of Maffucci syndrome have been published in the English literature. [1](#), [2](#) Note the image below.

Maffucci syndrome affects the skin and the skeletal systems. Superficial and deep venous malformations (hemangiomas) often protrude as soft nodules or tumors usually on the distal extremities, but they can appear anywhere. The hemangiomas are usually asymmetric. Venous-lymphatic malformations can occur but are much less common. Enchondromas are benign cartilaginous tumors that can appear anywhere, but they are usually found on the phalanges and the long bones. These bone abnormalities are usually asymmetric and cause secondary fractures. Approximately 30-37% of enchondromas can develop into a chondrosarcoma.

The hemangiomas in Maffucci syndrome manifest as blue subcutaneous nodules that can be emptied by pressure. The hemangiomas can be unilateral or bilateral and are usually asymmetric. Thrombi often form within vessels and develop into phleboliths. These phleboliths appear as calcified vessels under microscopic examination.

Enchondromas develop from the mesodermal dysplasia associated with Maffucci syndrome. As the bones grow, some cartilage material is left behind and grows irregularly, developing into the characteristic bone deformities. Bone irregularities in Maffucci syndrome include shortened length of the long bones, unequal leg length, pathologic fractures, and malunion of fractures.

In Maffucci syndrome, neoplastic changes occur in enchondromas. Chondrosarcoma is the most common neoplasm in this syndrome, affecting about 30% of patients. The average age for neoplastic change in Maffucci syndrome patients is 40 years. Vascular neoplasms have occurred in 4 reported cases: 2 hemangiosarcomas and 2 lymphangiosarcomas.

Maffucci syndrome is rare. Fewer than 100 cases of Maffucci syndrome have been reported in the United States.

Maffucci syndrome is rare, with about 160 total case reports in the English literature.

Usually in Maffucci syndrome, the skin and bone lesions progress slowly through the first or second decades of life. Bone and skin abnormalities cease by the second to third decade. Maffucci syndrome patients have a good prognosis if no malignant degeneration occurs. Patients usually have a normal life span.

No increased frequency of Maffucci syndrome occurs because of race.

Maffucci syndrome appears to be sporadically inherited. No sexual bias is present.

Lesions of Maffucci syndrome are first noted usually by age 4-5 years.

- Parents of a child with Maffucci syndrome first notice soft, blue-colored growths on the distal aspects of the extremities.
- Patients with Maffucci syndrome are usually short in stature and may have unequal arm or leg lengths due to the bone abnormalities.

## **Physical**

- In Maffucci syndrome, hemangiomas have been reported in various areas of the body, including the leptomeninges, the eyes, the pharynx, the tongue, the trachea, and the intestines.
- Enchondromas are usually found in the hands (89%), but they can be found on, although not limited to, the foot, the tibia, the fibula, the femur, the humerus, the ribs, and the skull. The tumors appear as nodular outgrowths and can cause a fracture, leading to further complications, such as shortened or unequal length limbs. Patients who are severely affected can have difficulty walking and manually manipulating objects. Note the images below.

The cause of Maffucci syndrome is unclear. Maffucci syndrome has no familial pattern of inheritance and appears sporadically

## **Treatment**

No medical care is needed in Maffucci syndrome patients who are asymptomatic. Patients do need careful follow-up care to evaluate any changes in the skin and bone lesions. The managing physician should arrange the proper consultations for the treatment of the patient.

- Radiologist: Radiography or CT scanning should be periodically performed to evaluate bone changes.
- Orthopedic surgeon: An orthopedic surgeon should be consulted to evaluate bone changes and skeletal neoplasms and to help in treating fractures associated with this disease.
- Dermatologist: A dermatologist should be consulted to help in evaluating hemangiomas associated with this syndrome and to discover any new lesions on the skin.

Physical activity is not limited for Maffucci syndrome. Some patients may have difficulty ambulating because of the bone abnormalities.