

## **Maffucci's syndrome**

Maffucci's syndrome, also known as: Kast's disease, Kast's syndrome, Maffucci-Kast syndrome.

### ***Synonyms:***

Achondromatosis with haemangiomas, chondrodysplasia angiomas syndrome, chondrodystrophy-haemangiomas syndrome; chondrodystrophy and vascular hamartoma syndrome, chondrodystrophy with angiomas, chondrodystrophy with vascular hamartoma, cutaneous dyschondroplasia-dyschromia syndrome, dyschondroplasia-angiomas syndrome, dyschondroplasia with haemangioma, dyschondrodysplasia-haemangiomas syndrome; multiple enchondromatosis syndrome; vascular hamartoma-dyschondroplasia syndrome.

### ***Description:***

Syndrome of enchondromas (benign tumours of cartilage), associated with multiple cavernous haemangiomas. Sometimes the patients show pigmentation. Normal at birth; bone and cartilage deformities appear during childhood in the years before puberty and the deformities increase during the period of growth. Complications are pathological fractures and other disorders of nonossified cartilage in the metaphyses and diaphyses of the long bones, chondrosarcoma and angiosarcoma. The skin and bony lesions are asymmetrical and do not coincide anatomically. Usually, no history of pain; orthostatic hypotension in sitting or standing position. Normal intelligence. Males are more frequently affected. Both sexes affected. Most cases are sporadic, but some instances of familial occurrence have been reported.

It was first described by Maffucci in 1881. In 1889, possibly unaware of Maffucci's article, Alfred Kast and Friedrich Daniel von Recklinghausen (1833-1910) reported another example of the same disorder. Thereafter the condition was known by a variety of descriptive titles and by the conjoined eponym, "Kast-Maffucci". However, Kast's name was soon abandoned and Maffucci's syndrome is now the accepted term. More than 100 hundred cases have been reported, including a series of 62 patients reviewed by Anderson in 1965.

The centenary of Maffucci's first description was commemorated in 1981 by Tilsley and Burden in a paper in which Maffucci's original captions were used in an account of a contemporary patient.

The Maffucci syndrome is closely related to Ollier's syndrome. See under Louis Xavier Édouard Léopold Ollier, French surgeon 1830-1900.

***Bibliography:***

A. MAFFUCCI, *Di un caso encondroma ed angioma multiplo. Contribuzione alla genesi embrionale dei tumori*, in «Il Movimento medico-chirurgico», Napoli, 1881, 3: 399-412 and 565-575.

A. KAST, *F.D. von Recklinghausen: ein Fall von Enchondrom mit ungewöhnlicher Multiplikation*, in «Virchows Archiv für pathologische Anatomie und Physiologie und für klinische Medicin», Berlin, 1889, 118: 1-18.

F. ANDERSON, *Maffucci's syndrome. Report of a case with a review of the literature*, in «South African Medical Journal», Cape Town, 1965, 39: 1066-1070.

D.A. TILSLEY, *P.W. Burden: a case of Maffucci's syndrome*, in «British Journal of Dermatology», Oxford, 1981, 105: 331-336.

Da: <http://www.whonamedit.com/synd.cfm/585.html> (consultata in rete il 1.2.2006).