

## Do you know this syndrome? *Você conhece esta síndrome?*

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### CASE REPORT

An 18-year-old white female patient went to the Orthopedics Department at the Hospital das Clínicas, in Belo Horizonte, complaining of pain in the right ankle since the age of six years, with progressive worsening. The pain had no relation with certain periods of the day and started after spraining the ankle.

Her past history included severe iron-deficiency anemia that required blood transfusion. She had no history of melena, hematochezia or hematemesis. The investigation of the gastrointestinal tract by means of endoscopy revealed esophagitis grade I of moderate intensity and erosive gastritis in the antrum. No vascular malformations were detected at colonoscopy.

The patient was referred to the Department of Dermatology to assess and treat angiomatic lesions that appeared at the age of four years and were stable.

On physical examination, the following changes were observed:

- Palpable nodules in the right proximal humerus, both iliac crests, proximal phalanges of

right second and third fingers; some nodules were painful at palpation;

- Enlarged right ankle;
- Presence of angiomatic nodules in the umbilical, right inframammary, left palmar and right plantar regions (Figures 1, 2, 3);
- Hyperchromic macule in the epigastric region;
- Deformities in right fingers (Figures 4 e 5).

The radiographs of ankles, hands, hip and humerus showed images suggesting synovial chondromatosis and enchondromas (Figure 5). Bone scintigraphy had findings suggesting enchondromatosis.

The patient was submitted to synovectomy in the right ankle, and hyperproliferative anterior medial synovium was identified during surgery. The pathological examination of the specimen demonstrated proliferation of irregularly arranged, tortuous and ectatic vascular channels, which were lined by epithelial cells with no atypia. The pathological diagnosis was "cavernous hemangioma".

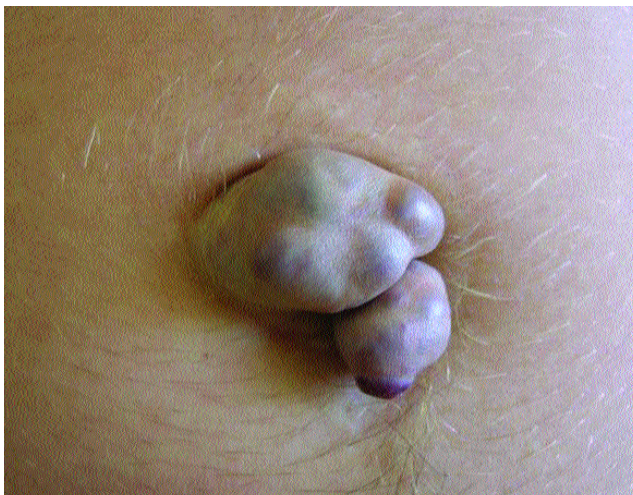


FIGURE 1: Angiomatic nodule in the umbilical region



FIGURE 2: Angiomatic nodule in the palmar region



FIGURE 3: Angiomatous nodule in the plantar region

### WHAT IS THIS SYNDROME?

#### Maffucci's syndrome

Maffucci's syndrome is a rare congenital mesodermal dysplasia, characterized by the association of cutaneous venous malformations with dyschondroplasia, which is a cartilaginous tissue dysplasia.<sup>1</sup> It shows no prevalence by sex or race.<sup>2</sup>

The skin lesions generally appear during childhood, by the age of four years; however, in 27% of patients they are present at birth.<sup>2,3</sup> The lesions proportionally grow with the individual and tend to have no spontaneous resolution. Lymphangiomas are often associated and may be the single cutaneous manifestation of the disease.<sup>1</sup> Other skin findings have been reported and include pigmentary changes, particularly café-au-lait spots.<sup>1,4</sup> The gastrointestinal tract



FIGURE 4: Deformities in right fingers due to bone alterations

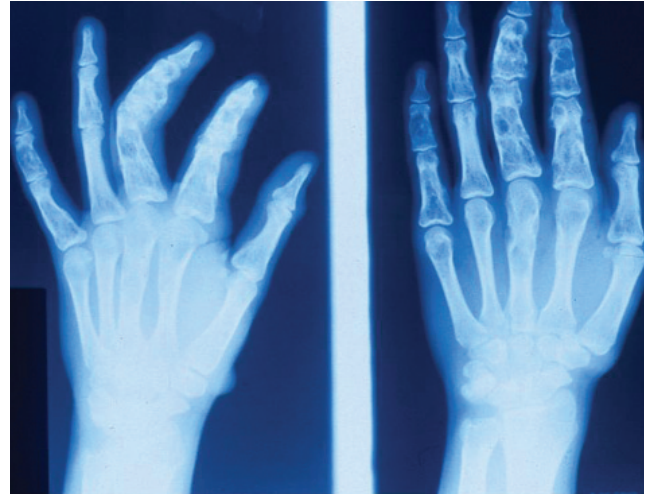


FIGURE 5: Radiograph of hands showing enchondromas

may present vascular anomalies.<sup>3,5-8</sup>

Enchondromas are benign cartilage-forming tumors revealing an osteolytic component at radiographs. They may be unilateral or asymmetric and involve mainly the phalanges, metacarpals, metatarsals and metaphysis of long bones.<sup>5,9-11</sup> The growth of affected bones is distorted and delayed due to interference in the epiphyseal cartilage, leading to pathologic fractures.

Several benign and malignant mesodermal tumors were reported in Maffucci's syndrome and the disease is potentially malignant. Chondrosarcoma is the most common malignant tumor and it may derive from enchondromas in 15% of cases. Other malignant mesodermal neoplasms could be associated, such as fibrosarcoma, angiosarcoma, lymphangiosarcoma, osteosarcoma and benign and malignant ovarian tumors. Non-mesodermal tumors, such as gliomas and pancreatic adenocarcinoma, were also described.<sup>2,5,10,11</sup>

The high malignant potential of the disease justifies the need for a careful follow-up of patients. Fast-growing or symptomatic lesions should be radiologically and histologically assessed. Some authors suggest periodical imaging studies of the abdomen, pelvis and central nervous system for screening.<sup>3</sup>

The surgical treatment of skin lesions should be performed for esthetic reasons.<sup>1</sup> □

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