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Case Report

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Chondrosarcoma of the Humerus Complicating Maffucci Syndrome

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Summary

Maffucci syndrome is a dysplastic condition of the mesoderm, characterized by the combination of multiple enchondromes and skin or visceral angiomas. It is a very rare non-hereditary congenital disease. Ollier's disease is characterized by multiple enchondromes without benign vascular overgrowth as in Maffucci syndrome. The risk of sarcomatous malignant degeneration is common in both pathologies. We report a case of malignant transformation into chondrosarcoma of the left humerus in a forty-year-old patient followed for Maffucci syndrome.

Keywords: *Maffucci syndrome; chondrosarcoma.*

Introduction

Maffucci syndrome is a mesodermal dysembryoplasia which associates benign cartilaginous tumors (enchondromas) which mainly sit in the phalanges and long bones. The transformation of chondromas into sarcomas is frequent in 15 to 30% of cases (1,2). Sarcomas preferentially affect lesions on the pelvis and femur. The onset of a periosteal reaction, cortical osteolysis and invasion of the soft tissues are the main signs suggesting malignant transformation into chondrosarcoma (3).

Observation

This is a forty-year-old woman, the first of a sibling of six children with the notion of first degree consanguinity. She says she underwent surgery on her left leg at the age of 8, the indication of which is not well known. She consulted with inflammatory left shoulder pain that had progressed for three months. The clinical examination found a patient of small size (1.41meter), an inequality of the lower limbs, a left genu varum and skin nodules of soft consistency, bluish and not painful in the lower limbs (figure 1a) suggesting a hemangioma during pathological examination. Hard swelling was found on the fingers (figure 1b). The left shoulder was swollen, red, hot on palpation and limited to mobilization (figure 1c).

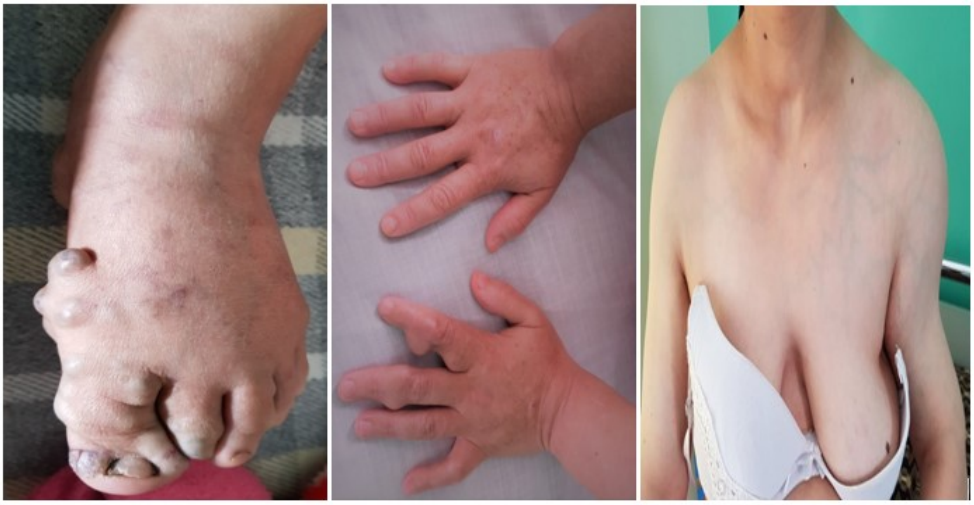

Figure 1a: Skin nodules of soft consistency, bluish and painless (hemangioma). **Figure 1b:** hard swelling on the fingers. 1c- Left shoulder swollen, red, hot on palpation and limited to mobilization.

Blood analysis showed: biological inflammatory syndrome (ESR = 33mm, CRP = 46mg / l) blood count, TSH, PTH, calcium levels were normal. The standard radiograph showed a blown appearance with rupture of the cortex of the left humeral head (figure 2a); and other lesions reminiscent of enchondromas, found in the hip, knee and left ankle. A positron emission computed tomography scan taken two years earlier revealed the presence of a large mass in the moderately hyper-metabolic left shoulder, destroying the left humeral head and extending outward to the adjacent muscular and subcutaneous planes, suspected of 'degeneration (figure 2b).

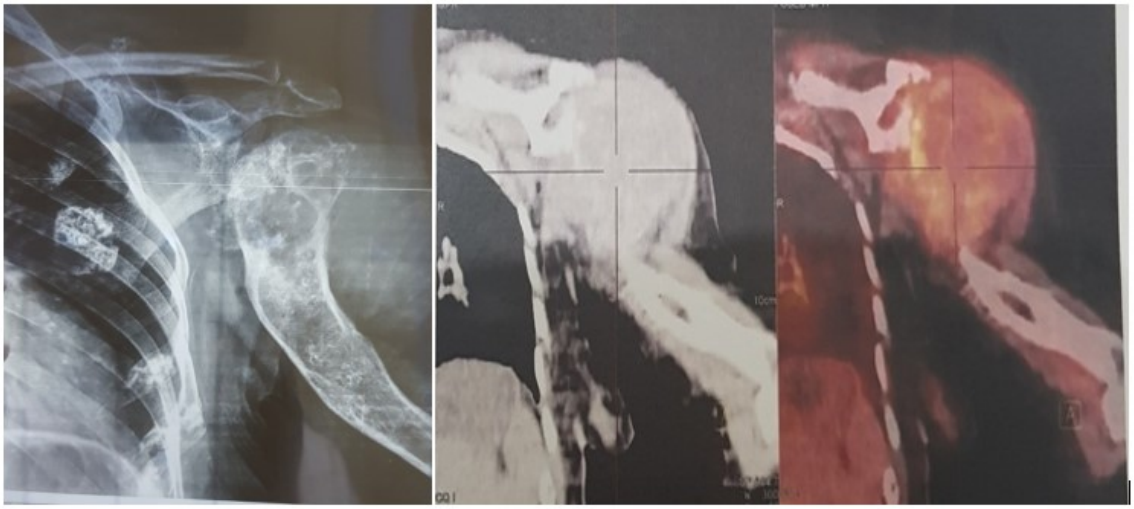

Figure: 2a: X-rays showing a blown appearance of the cortical humerus with cortical lysis. **Figure 2b:** PET CT showing a large mass of the hypermetbolic shoulder destroying the humeral head extending outwards towards the muscular and skin planes.

The anatomopathological study after surgical biopsy of the soft part of the shoulder revealed a morphological appearance compatible with grade 2 chondrosarcoma. Radical surgical treatment by inter-scapulothoracic disarticulation offered to the patient was refused. Lost to follow-up, she consulted two years later with MRI of the left shoulder which revealed an aspect compatible with a sarcomatous tumor of the left arm with a probable muscular starting point with extensive bone invasion infiltrating the brachial plexus without real sheathing, associated with secondary bone and pulmonary lesions (figure 3).

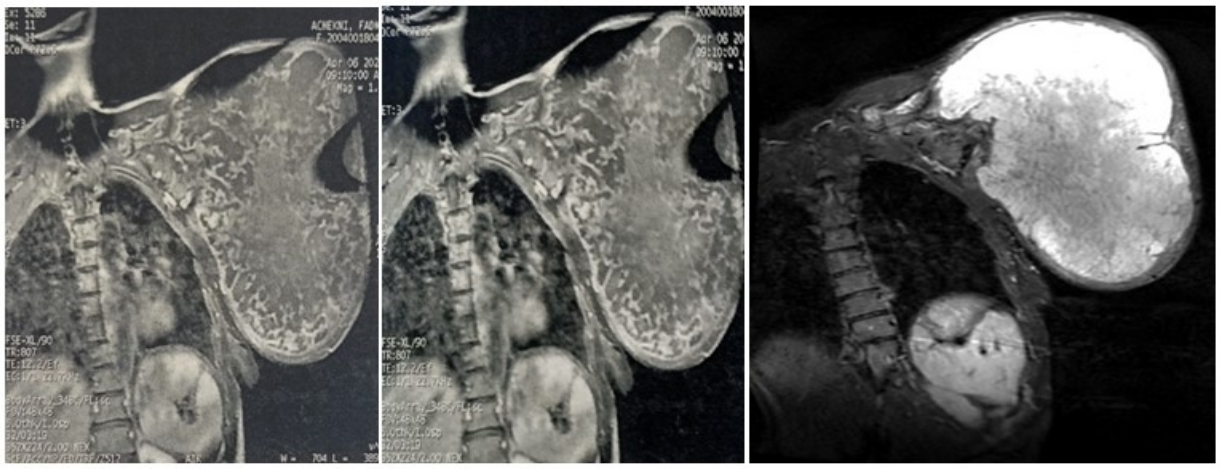

Figure 3: Large tissue process of the upper 2/3 of, osteolytic, largely necrotic, in T1 hypointense and T2 hypersignal enhanced heterogeneously after injection of gadolinium, site of calcifications.

Complementary CT scan of the thorax revealed multiple parenchymal nodules and micronodules, a frosted glass appearance, and staged osteolytic lesions of the ribs and left scapula of secondary origin.

Discussion

Maffucci syndrome is a rare disease. More than 200 cases have been reported in the literature [4,5] and to our knowledge 5 cases in Morocco [1,6]. This dyschondroplasia usually begins between 1 and 5 years old. Lewis and Ketchum in their extensive review of the world literature on 105 cases found that clinical manifestations are present in 25% of cases at birth or in the first year, in 45% before the age of 6 years and in 78% before puberty [7]. The first sign in our patient was noted at the age of 8 years.

This syndrome is associated with an increased risk of malignant degeneration preferentially concerning lesions located on the pelvis and the femur. The transformation of chondromas into sarcoma is the most frequent, 15 to 30% of cases [1,8]. Chondromas develop in long and short bones, with a predilection for the phalanges, pastern, and long bones of the legs and forearms [9]. In a study conducted to

better understand the natural history of Ollier's disease and Maffucci syndrome. 287 articles describing these violations had been examined and surveys had been distributed directly to 162 patients via Facebook. Vascular abnormalities were identified only in internal organs in four patients; and the prevalence of cancer in affected patients was about 50%. [10] In our patient the internal organs explored were normal. A similar case was reported by García-Ortega DY et al. in a 44-year-old man where high-grade multicentric fuso-cellular chondrosarcoma and sarcoma of the scapular and tricipital region was treated by inter-scapulothoracic disarticulation. The course was marked by pulmonary metastasis despite radical treatment. There is the problem of delay in taking charge. Jurik AG et al offers magnetic resonance imaging as a screening method for detecting malignant transformation in patients with Maffucci syndrome. [11] The treatment remains disappointing, being limited to analgesics and surgical interventions for tumor resection and correction of deformity [12].

Conclusion

The swelling and the appearance of soft tissue pain in a patient followed for Maffucci syndrome should suggest sarcomatous malignant degeneration. The appearance of cortical osteolysis, a periosteal reaction and invasion of the soft tissue are the main signs in favor of this transformation. Treatment is limited to analgesics and surgery for tumor resection and deformity correction. Regular radio-clinical monitoring is necessary. MRI remains the test of choice for screening for malignant transformation.

Declaration of links of interest

The authors declare that they have no competing interest.

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