

HAND INVOLVEMENT IN OLLIER DISEASE AND MAFFUCCI SYNDROME: A CASE SERIES

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Abstract

Ollier Disease and Maffucci Syndrome are two rare diseases that can cause tumors in several organs, having a special predilection for the hand. However, there have been very few reports in the literature focusing on hand manifestations of these diseases.

We report the cases of three female patients: one with Ollier Disease, and two other with Maffucci Syndrome. All patients had hand involvement as their initial primary complaint. The Ollier Disease patient developed chondrosarcomas of two digits and had to have these fingers amputated. One of the Maffucci patients died one year after presentation from a brain glioblastoma.

These cases emphasize the importance of early diagnosis of Ollier Disease and Maffucci Syndrome, as these two conditions are associated not only to crippling hand deformity, but also to a significant risk of chondrosarcoma, and other malignant tumors.

Keywords: Ollier Disease; Multiple Enchondromatosis; Chondrosarcoma; Maffucci Syndrome; Hand.

Introduction

First described by Ollier in 1900¹, Ollier disease is a rare, nonfamilial disorder characterized by multiple enchondromas and areas of dysplastic cartilage²⁻³. The estimated prevalence of Ollier disease is one in 100 000⁴. Maffucci syndrome is an even rarer condition that was described for the first time

in 1881⁵, being characterized by the association of enchondromatosis and hemangiomas⁶. Hemangiomas can affect skin, mucosal surfaces and internal organs⁷. There are only slightly over of 200 cases of Maffucci syndrome reported in the literature worldwide⁸⁻⁹.

The ability to identify these two diseases is crucial, as they are associated with a significant risk of chondrosarcomas and other malignant tumors, that have a better prognosis if treated early⁴.

Case Series

Case 1

A 48-year-old right-handed female appointed at the Department of Plastic Surgery at our hospital complaining of functional and aesthetic problems with her left hand and foot caused by soft tissue swellings since she was 14 years old. She mentioned that the lesions in the third and fourth digits of her left hand had started to increase in size in the previous month and that, since then, she suffered from an ever growing pain in those digits (Figure 1A).

Radiographs of the hand revealed features suggestive of enchondromas, but with signs of possible malignant transformation (Figure 1B). The diagnosis of Ollier disease was established. Biopsies of the finger nodules were performed, revealing only evidence of enchondromas. As the patient continued to complain of pain, the nodules were excised, preserving the fingers (Figure 2A). However, as histopathological analysis of the nodules revealed chondrosarcomas in both fingers, amputation of the third and fourth digits was performed. Further histological analysis of the amputated fingers revealed complete excision of the tumors. One year after surgery, the patient showed no signs of recurrence and had acceptable left hand function (Figure 2B).

Case 2

A 33-year-old right-handed female came to the

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Figure 1. Patient with Ollier disease and signs of malignant transformation
A photograph of the hands (**1A**) shows large subcutaneous nodules in the third and fourth fingers of the left hand, corresponding to enchondromas.
A radiograph of the left hand (**1B**) shows, at the base of the proximal phalanx of the second finger, an oval, radiolucent, and homogenous lesion with a well-defined bony margin. This radiographic presentation is highly suggestive of enchondroma. In contrast, at the level of the third and fourth digits, there are multiple cystic-like lesions, with various radiographic signs of malignant transformation, namely: cortical bone erosion, extension of the tumor into the soft tissues, margin irregularity and uneven pattern of mineralization^{2,5}

Hand Clinic complaining of insidious growth of several small nodules in her hands since she was 13 years old. In the previous fifteen years she also noticed the progressive development of areas of bluish discoloration scattered through her upper limbs that bleached with pressure (Figures 3A and 3B). Radiographs of the hands revealed the presence of multiple enchondromas and phlebolitis (Figure 3C). The patient was diagnosed with Maffucci syndrome. Even though surgery was offered to excise some of the enchondromas, and several options were presented to treat the hemangiomas,

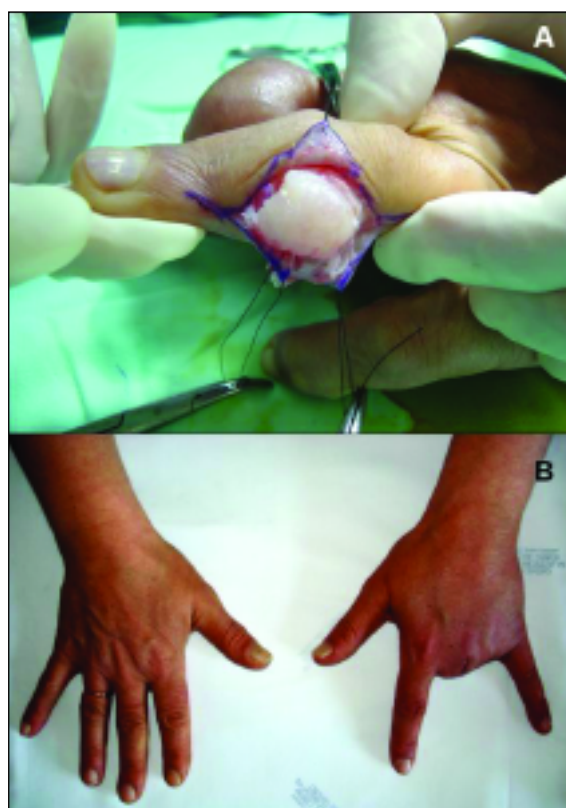


Figure 2. Treatment of a patient with Ollier disease
(**2A**) Surgical dissection of the nodule in the third finger of the left hand of the patient depicted in Figure 1 reveals a light-grey oval-shaped cartilaginous lesion within the osseous portion of the middle phalanx
(**2B**) As the nodules in the third and fourth fingers proved to be chondrosarcomas, proximal digital amputation was performed. One year after amputation, the patient had a reasonable hand function

the patient declined any procedure for the time being, and continues to be followed regularly in the Hand Clinic.

Case 3

A 28-year-old right-handed female was referred to the Hand Clinic because of multiple continuously growing nodules in both her hands since she was 8 years old (Figures 4A and 4B). The patient also presented multiple hemangiomas all over her body since that time. She was diagnosed with Maffucci syndrome. An arteriography of both her hands was performed (Figure 4C) revealing the classical pattern of hand hemangiomas. She was submitted to excision of the nodules that had been growing most rapidly. Microscopical analysis of the surgical specimens was consistent with the diagnosis of en-



Figure 3. Hands of a patient with a moderate form of Maffucci Syndrome

Photographs of the palmar (A) and dorsal (B) regions show multiple hard subcutaneous nodules in both forearms and hands, compatible with multiple enchondromas. There are also several areas of bluish discoloration consistent with hemangiomas. Radiograph of the hands (C) shows multiple small, radiolucent, and homogenous lesions, with a well-defined bony boundary visible, corresponding to enchondromas. In addition, there are multiple small round calcifications in the surrounding soft tissues, suggestive of phleboliths, which are typical of cavernous hemangiomas

chondromas. One year after her initial consultation, she died of a rapidly expanding brain glioblastoma.

Discussion

Ollier disease and Maffucci syndrome are two rare diseases that can be diagnosed relatively easily solely on clinical grounds and taking into account a few simple ancillary tests, namely radiographs and, occasionally, arteriography^{2,4}.

In these diseases, the most frequent locations of enchondromas are the small bones of the hands and feet, the long tubular bones, and also the flat bones like the pelvis^{2,4}. The trunk is usually spared^{2,4,6}.

Even though Ollier disease and Maffucci syndrome have been said to be two manifestations of the

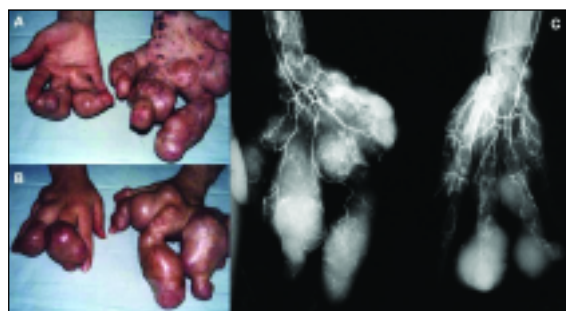


Figure 4. Hands of a patient with an extreme form of Maffucci Syndrome

Photographs of the palmar (A) and dorsal (B) regions show multiple large enchondromas in both hands, which not only cause great deformity, but also significantly hamper the function of both the involved segments and of the uninvolved digits close to them. In the left hand, subcutaneous vessels are dilated and tortuous, and several hemangiomas are visible.

Hand arteriography (C) shows anomalous hand vasculature in both hands. Terminal vessels originate a fine mesh of small vessels that supply round or ovoid areas of hypervascularity. These represent hemangiomas. Multiple lytic lesions are distributed centrally and eccentrically in the phalanges and metacarpal bones of both hands, representing enchondromas

same mesenchymal dysplasia¹⁰, it is of clinical interest to differentiate the two, as the risk of sarcomatous change is much higher in patients with Maffucci syndrome, ranging from 17% to 50% of the cases to an astounding 100% after prolonged follow-up². In Ollier disease, on the contrary, sarcomatous transformation occurs only in 25–30% of cases¹¹.

Skeletal lesions that enlarge or become painful without prior trauma are suspected of having undergone malignant degeneration and need to be biopsied without delay². Similarly, suspicious radiological lesions, should merit prompt biopsy⁴.

In the absence of clinical symptoms or problems, no treatment is needed^{2,4}. Surgery is indicated only in the case of complications, such as pathological fractures, growth defects and malignant transformation^{2,4}. The goals of surgery are to remove the tumor mass and to allow the histological diagnosis. This could reveal lesions possibly requiring adjunctive therapies (e.g., chondrosarcomas)². Sclerotherapy, irradiation, laser therapy and surgery for the vascular lesions in Maffucci syndrome have also been described⁸.

In conclusion, Ollier disease and Maffucci syndrome are two rare diseases whose hand ma-

nifestations are considered pathognomonic¹¹. Their timely diagnosis is of paramount importance, as these two conditions are associated not only to crippling hand deformity, but also to a significant risk of chondrosarcoma, and other malignant tumors⁴.

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