CASE CLÍNICO

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...
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 phlebolits (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, 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-
same mesenchymal dysplasia, 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-up. In Ollier disease, on the contrary, sarcomatous transformation occurs only in 25–30% of cases.

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

In the absence of clinical symptoms or problems, no treatment is needed. Surgery is indicated only in the case of complications, such as pathological fractures, growth defects and malignant transformation. 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). Sclerotherapy, irradiation, laser therapy and surgery for the vascular lesions in Maffucci syndrome have also been described.

In conclusion, Ollier disease and Maffucci syndrome are two rare diseases whose hand involvement is quite common. 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.

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. The trunk is usually spared.

Even though Ollier disease and Maffucci syndrome have been said to be two manifestations of the same mesenchymal dysplasia, 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-up. In Ollier disease, on the contrary, sarcomatous transformation occurs only in 25–30% of cases.

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In conclusion, Ollier disease and Maffucci syndrome are two rare diseases whose hand ma-
manifestations are considered pathognomonic. 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 tumors.

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References

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