Medial canthal lesions in a man with a scarred wrist: Subcutaneous lymphangiomas in association with Maffucci’s syndrome

J. Godfrey Heathcote, MB, PhD, FRCP,1 Alejandra A. Valenzuela, MD2

1. Professor of Pathology and Ophthalmology, Pathology Department, Faculty of Medicine, Dalhousie University, Nova Scotia, Canada
2. Associate Professor, The George Haik Sr./St. Giles Foundation Chair, Ocular & Lacrimal Diseases/Surgery, Oculoplastics & Periocular Eyelid Reconstructive Surgery, Orbital & Ocular Adnexal Oncology, Department of Ophthalmology, Tulane University Medical Sciences Centre, School of Medicine

Financial/proprietary interest: None

Abstract

A case of Ollier’s disease is presented, in a male adult with multiple enchondromas. The patient was reclassified as a Maffucci lymphangioma syndrome after two periorbital lymphangiomas were surgically excised.

Key words: Maffucci syndrome; hemangiomas, periorbital disease.

Introduction

Maffucci syndrome is distinguished by multiple soft tissue hemangiomas and multiple enchondromas. Ollier disease includes enchondromatosis without hemangiomas and these two conditions are now thought to represent a spectrum of congenital mesenchymal dysplasia. We present a case of a 49 year-old male affected by multiple enchondromas, who was reclassified as a Maffucci lymphangioma syndrome after two periorbital lymphangiomas were surgically excised.

Case report

A 49-year-old man was referred because of two right medial canthal lesions, one above and one below the canthus. They had been present for two years and intermittently swelled. On examination, both lesions appeared to be well-defined, the superior more adherent to deep structures and felt cystic on palpation, while the inferior was more superficial and appeared more vascular. Visual acuities were 20/25 OU with otherwise normal ocular findings. CT scan confirmed the well-circumscribed lesions, which enhanced with contrast, showing no calcification or bony defects (Figure 1). Both lesions were excised in toto and submitted for histopathological examination (Figure 2). At the time of consultation, the patient had a large scar on his left wrist. On enquiry, he indicated that, at the age of 16 years, he had sustained a fracture of the femur and a diagnosis of Ollier’s disease had been made. Two tumors had been removed from the femur one year later and a further enchondroma removed from the right tibia at the age of 29 years.

Corresponding author:
Alejandra A. Valenzuela MD
1430 Tulane Avenue, Sl-69 New Orleans, LA, 70112
Office phone: (504) 988-2465, Office fax: (504) 988-2684
avalenz1@tulane.edu
Date of submission: 22/11/2012 Date of approval: 22/01/2013

Fig. 1: Axial CT scan showing a well-circumscribed 1x1cm lesion in the left superomedial orbit, which enhanced with contrast. The mass appeared to be in close relation to the superior globe and does not show calcification or induces a bony defect.
At the age of 44 years, a cartilaginous tumor was removed from the left distal radius. This was initially diagnosed as an enchondroma but one year later it recurred and was completely excised: the final pathological diagnosis was chondrosarcoma grade 1. Three years before his current problem, he had developed a soft tissue swelling in the front of the right tibia. This was considered to have resulted from spillage of the enchondroma removed previously from this site but on excision was found to be a cavernous hemangioma. The diagnosis of Ollier’s disease was modified to Maffucci’s syndrome. On histopathology, both specimens consisted of rubbery tan tissue measuring 7 and 9mm in maximal dimension and their microscopic appearance was similar. Numerous vascular spaces lined by endothelial cells were seen within fibromuscular tissue. Some spaces were linear, but others and rounded profiles. They did not contain significant amounts of blood, but some did contain wispy proteinaceous material, consistent with lymphatics. Some of the endothelial cells were flat; others had plump nuclei but these were not atypical and no mitotic activity was observed. There was not any smooth muscle in the vessel walls. Immunoperoxidase studies indicated that the endothelial cells expressed CD 31, Factor VIII and D2-40, the latter being a marker of lymphatic endothelium. Spindle cells were not seen in the stroma, but there were infiltrates of lymphocytes and plasma cells. Peripheral nerve bundles were a prominent feature of the surrounding tissue. The vascular lesion was felt to be a lymphangioma, arising in the contest of Maffucci’s syndrome. The lesion extended to the margin of excision and follow-up for recurrent disease was suggested.

**Discussion**

In 1881, Maffucci described a condition characterized by multiple soft tissue hemangiomas and multiple enchondromas. Ollier in 1900 described enchondromatosis without hemangiomas and the two conditions are now thought to represent a spectrum
of congenital mesenchymal dysplasia. Maffucci's syndrome is rare, congenital and non-hereditary and shows no racial or gender predilection. No consistent genetic abnormality has been identified. In addition to the vascular and cartilaginous tumors, patients show a propensity to develop other mesenchymal, e.g., fibrous dysplasia, and nonmesenchymal, e.g., glioma, tumours.

The vascular lesions are often present at birth, large in proportion to body size and, unlike infantile hemangiomas, do not regress. They include spindle cell hemangiendothelioma, probably the commonest, and cavernous hemangioma, many of which are now thought to be spindle cell hemangiendotheliomas. In addition, capillary hemangiomas, arterio-venous malformations and lymphangiomas have been described. Recently composite hemangiendothelioma, a locally aggressive neoplasm, with a combination of benign, malignant and intermediate vascular elements has been linked to the syndrome. The enchondromas generally manifest before puberty and are predominantly unilateral, occurring particularly in the long bones of the arms and legs and the phalanges. In the skull they are generally in the sphenoid bone.

The enchondromas contain areas of dysplastic cartilage, characterized by hypercellularity and the presence of binucleate chondrocytes with visible nucleoli. The distinction from a low-grade chondrosarcoma may be very difficult and behavior is an important indicator of malignant transformation. Permeation between existing bony trabeculae is seen as one of the more reliable histological sings of malignant transformation. Malignant transformation is accompanied by loss of heterozygosity at chromosomal sites 13q14 and 9p21, perhaps indicative of loss of tumor suppressor genes, and over expression of p53. Lymphangiomas have been reported in Maffucci's syndrome and Auyeung et al. proposed that a Maffucci lymphangioma syndrome should be considered a variant of Ollier's disease. He reviewed the cases of seven patients from the literature: 2/7 showed lymphangiomas only; 5/7 had lymphangiomas with other vascular lesions; 5/7 were women and all had enchondromas of the axial skeleton. The pathogenetic link between enchondromatosis and vascular malformations has not been uncovered. Robinson et al. in 1994 postulated an underlying neural abnormality with increased numbers of nerves in the malformation and in the enchondromas. These nerves were judged to be more active, based on the expression of calcitonin gene related peptide, a known mitogen for mesenchymal cells.

More recently, increased numbers of small and large nerves have been shown in vascular malformations of different types, particularly in the head and neck region. Increased numbers of nerves were observed in 87% of AV malformations, 55% of venous malformations and 36% of lymphatic-venous malformations (but only in the venous component). Among the ophthalmic manifestations of Maffucci’s syndrome, the effects of intracranial astrocytomas and cranial nerve palsies as a result of skull base tumors have figured prominently. Multiple, bilateral intra-orbital hemangiomas have also been reported, although the lesions were not confirmed by histopathology.

**REFERENCES**