OCULAR FINDINGS IN CARNEY COMPLEX

MANIFESTACIONES OFTALMOLÓGICAS DEL SÍNDROME DE CARNEY

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ABSTRACT

Case report: Carney complex (CNC) is a multiple neoplasia syndrome, inherited in an autosomal dominant manner, characterized by myxomas, spotty skin pigmentation, endocrine tumors and ophthalmic abnormalities.

Discussion: We report the ocular findings which usually precede the most serious component of the complex, the cardiac myxoma (Arch Soc Esp Oftalmol 2006; 81: 709-712).

Key words: Carney complex, pigmentation disorders, myxomas, endocrine gland neoplasms.

INTRODUCTION

Carney’s Syndrome or Complex (CNC) was first described by J.A. Carney in 1985. It is a multineoplastic syndrome associating cardiac, endocrine, cutaneous and neural tumors as well as a variety of skin and mucous pigmented lesions. Most of the cases are hereditary (70%) but the literature has also documented some sporadic cases under the acronym of LAMB (Lentigines, Atrial Myxomas and Blue Nevi) and NAME (Nevi, Atrial Myxomas and Ephelides). CNC is a dominant autosomic gene exhibiting a variable penetration and expression, and is linked to two loci: 17q22-24 and 2p16. All over the world about 300-500 patients are known to have this syndrome (1).

CASE REPORT

A 36-year old woman recently diagnosed with CNC who visited our practice due to non-specific...
ocular irritations in her left eye (LE) with a 9-month evolution. She did not exhibit family history for CNC or other tumors.

The general expressions are lentiginous lesions in the skin of the face and breasts, spongy subcutaneous nodules at the cervical level, melanocytic psammomatose Schwannoma, GH-producing hypophysitarian microadenoma and bi-auricular mixomes (fig. 1), all diagnosed in the past 2 years, although the patient started exhibiting symptoms 5 years ago.

The only ophthalmological history the patient exhibited is the removal of a small tumoration in the left palpebral free edge 3 years ago, with microscopic diagnostic of myxomatous cutaneous tumor. The ophthalmological exploration revealed the following:

— Pigmented lesion in left perilimbar temporal conjunctiva, 4 mm diameter, suggesting conjunctival compound lentigo or nevus with a 2-year evolution (fig. 2). Controlled only with regular assessments.

— Small rounded myxomatous lesion in the right periorocular skin in the form of a smooth subcutaneous nodule which did not change in the past years (fig. 3).

— Papillomatous tumor in the LE inferior palpebral free edge with benign characteristics (fig. 2). This tumor was removed for the second time and informed with the same microscopic characteristics of the first one.

**DISCUSSION**

The clinical expressions of this syndrome are as follows: 1. Lentiginosis, dotted skin pigmentation with typical distribution (lips, conjunctiva and

Fig. 2: Flat lentiginous pigmented lesion in perlimbar conjunctiva. Myxomatous tumoration in left inferior palpebral free edge.


For diagnosing this pathology it is necessary to present either two clinical expressions or a clinical
one and supplementary criteria. These are: first order relative affected or mutation deactivating gene 17q22-24.

Morbidity and mortality are related to cardiac myxoma, the second most common component of this syndrome. This tumor can cause sudden death in up to 16% of cases (1). The endocrine tumors associated to this complex can cause high morbidity, together with some malign tumors which could be fatal such as thyroid nodules or melanocytic Schwannomae.

Skin alterations are most frequent, such as lentiginous lesions and blue nevus, primarily located on the face, eyelids, ears and edge of lips.

Cutaneous multiple myxomas are also frequent and recur after excision (2). The most frequent locations thereof are the eyelids, genitals and hearing channel. Upon finding a superficial angiomixome it is recommended to search for other components of this disease (1).

The ophthalmic alterations which may occur are (in order of frequency): facial and palpebral lentigous, pigmented lesions in the caruncle and palpebral myxomas (3). The literature has described cases of plexiform pigmented Schwannomas in the uvea (4).

The differential diagnostic of pigmented lesions in the caruncle or conjunctiva includes melanocytic nevus, congenital or acquired melanosis, malign melanoma and pigmentation secondary to drugs.

In our case it must be emphasized that the palpebral myxoma went unnoticed (in the ocular assessment) as a member of this disease, while probably being the earliest component thereof. The patient did not return to the ophthalmic checkup until after 3 years, and she only did so due to a relapse of the myxoma but she never saw an ophthalmologist for the asymptomatic conjunctival pigmentation.

Accordingly, as the aforementioned ocular symptoms (which in turn are part of major clinical criteria) usually precede the other, more serious, findings of the syndrome, it is very important to obtain an early identification and diagnostic by the ophthalmologist (5). We can help to diagnose and treat the fearful cardiac pathology before it becomes symptomatic. Annual echocardiographic controls must be made. In addition, the endocrine and gonadal conditions associated to this syndrome must also be screened.

**REFERENCES**