Familial Presentation of Multiple Scalp Tumors

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Abstract. A 74-year old woman was referred for evaluation and treatment of a tumor measuring 3 × 2 cm in the left preauricular region that has been progressively growing in the past years. Physical examination revealed an hemispheric and firm tumor with reddish coloration and telangiectases. The patient has been using a wig during the past 15 years to cover the scalp lesions. The patient underwent complete surgical excision of the scalp and the defect was reconstructed with partial-thickness grafts.

Given the long-standing evolution and extension of the tumors involving the scalp and face we suspected a familial cylindromatosis syndrome therefore, we revised her six children observing scalp tumors in two daughters. The tumors were excised and the histological findings confirmed the diagnosis of cylindromas.

Key words: multiple cylindromatosis, Brooke-Spiegler syndrome, Poncet-Spiegler cylindroma, familial cylindromatosis or turban tumour syndrome.

Case Description

A 74-year old woman was referred for evaluation and treatment of a tumor measuring 3 by 2 cm in the left preauricular area, which had been growing progressively in recent years. Physical examination revealed a clearly delimited hemispheric, hard tumor, with reddish coloration and telangiectases (Figure 1).

Figure 1. (Preoperative anterior view.) On examination a clearly delimited hemispheric, firm, rounded tumor was observed, with reddish coloration and telangiectasias.
The patient wore a wig which she removed for a peripheral examination of the lesion to reveal the presence of multiple tumors of varying sizes distributed all over the scalp. These were nodular, of a firm consistency, and generally of the same characteristics as the first tumor examined. The larger tumors were deformed and ulcerated.

Following surgical treatment of the lesions, all of the woman’s descendents were contacted for an appointment and examined for similar lesions. Two of them presented small tumors.

The patient underwent total excision of the scalp to the level of the galea aponeurotica, followed by reconstruction with partial-thickness grafts.

Given the extensive distribution of tumors across the scalp and face and the long-term evolution of the condition, we suspected familial cylindromatosis. We therefore examined her 6 children, observing tumors on the scalp of 2 of them, and these were removed. Histological findings confirmed the diagnosis of cylindromas.

Discussion

Multiple cylindromatosis, also known as Brooke-Spiegler syndrome, Poncet-Spiegler cylindroma, familial cylindromatosis, or turban tumor syndrome, is a benign neoplasm of autosomal dominant inheritance, characterized by the development of multiple tumors such as cylindromas, trichoepitheliomas, and occasionally spiradenomas. In terms of etiology, mutations have been described in association with the CYLD gene (16q12-13 for cylindroma and 9p21 for trichoepithelioma).

Cylindromas are epithelial tumors, specifically of the skin appendages (apocrine glands and pilosebaceous units), which occur from puberty and throughout adult life, predominantly in women, with a predilection for the scalp. The tumors tend to occur in isolation, but a case of Brooke-Spiegler syndrome like the present one is characterized by multiple, hemispherical, clearly delimited, excrecent nodules of a variable size, firm consistency, and reddish color. The number and (slow) growth of these can result in considerable disfigurement.
Conflicts of interest
The authors declare no conflicts of interest

References