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CASE REPORTS

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.

TUMORACIONES MÙLTIPLES EN CUERO CABELLUDO DE PRESENTACIÒN FAMILIAR

Resumen. Se trata de una mujer de 74 años, que acude a la consulta remitida para valoración y tratamiento de tumoración de 3 × 2 cm en región preauricular izquierda, que había ido creciendo progresivamente a lo largo de los últimos años. A la exploración, se observa un tumor duro, globuloso, de carácter hemisférico, de bordes bien delimitados, de color rojizo con telangiectasias. La paciente usaba peluca desde hace quince años para ocultar las lesiones de su cuero cabelludo.

La paciente fue intervenida realizando exéresis completa del cuero cabelludo hasta el plano de la galea, mientras que la reconstrucción se realizó con injertos cutáneos de espesor parcial.

Dada la extensión de las tumoraciones, generalizadas por el cuero cabelludo y la cara y su larga evolución, sospechamos un síndrome de cilindromatosis familiar, por lo que citamos a consulta a sus seis hijos, encontrando en dos de ellos tumores en cuero cabelludo que fueron extirpados. El resultando de la anatomía patológica fue en los dos casos de cilindromas.

Palabras clave: cilindromatosis múltiple, síndrome de Brooke-Spiegler, síndrome de los cilindromas de Poncet-Spiegler, cilindromatosis familiar o síndrome del tumor en turbante.

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).

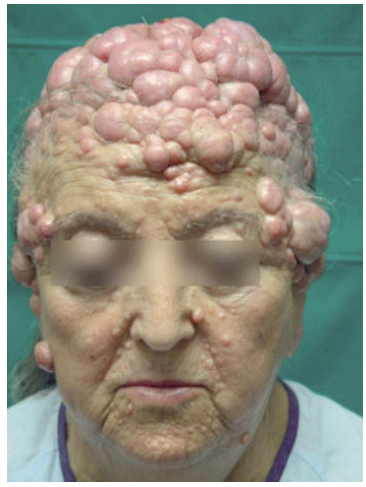

Figure 1. (Preoperative anterior view.) On examination a clearly delimited hemispheric, firm, rounded tumor was observed, with reddish coloration and telangiectasis.

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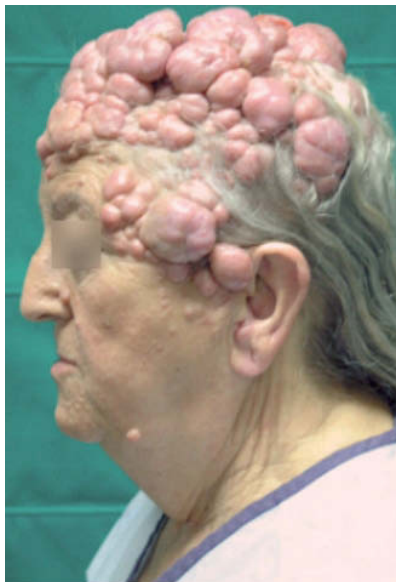

Figure 2. (Lateral view.) The patient wore a wig which she removed for a peripheral examination of the lesion to reveal the presence of multiple tumors of varying sizes distributed all over the scalp. These were nodular, of a firm consistency, and generally of the same characteristics as the first tumor examined. The larger tumors were deformed and ulcerated.

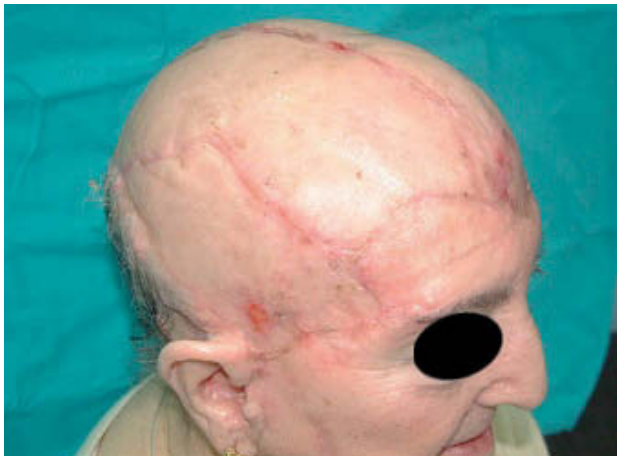

Figure 3. At 3 months after the operation. Partial-thickness skin grafts.

The patient wore a wig which she removed for a peripheral examination of the lesion, revealing the presence of multiple tumors of varying sizes distributed all over the scalp. These were nodular, of a firm consistency, and generally showed the same characteristics as the tumor examined initially, although the larger tumors showed deformation and ulceration. The patient reported that these had been developing for 15 years (Figure 2).

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 (Figure 3).

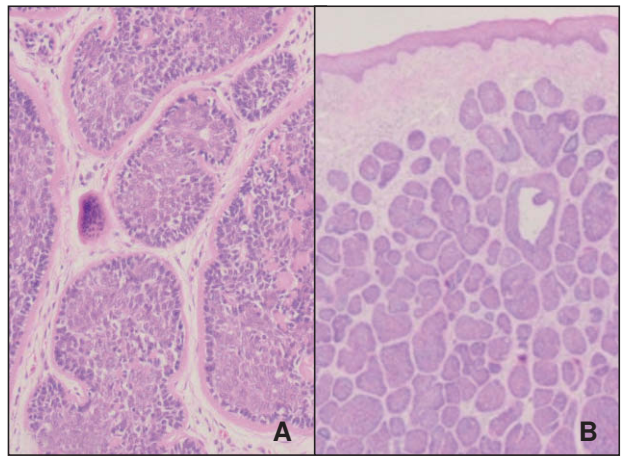

Figure 4. A. Poorly defined cutaneous tumor, comprising nests and fascicles of basaloid cells surrounded by an eosinophilic hyaline band (hematoxylin-eosin, ×2).
B. At a higher magnification a double population is seen: peripheral cells with a hyperchromatic nucleus and a tendency to palisade and other larger cells with a vesiculous central nucleus (hematoxylin-eosin, ×20).

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 (Figure 4).

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.[1-6] In terms of etiology, mutations have been described in association with the CYLD gene (16q12-13 for cylindroma and 9p21 for trichoepithelioma).[7-14]

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, excrescent nodules of a variable size, firm consistency, and reddish color. The number and (slow) growth of these can result in considerable disfigurement.[15-19]

Conflicts of interest
The authors declare no conflicts of interest

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