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Collision tumor in Brooke-Spiegler syndrome: a case report

Tumor de colisión en el síndrome de Brooke-Spiegler: reporte de un caso

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None.

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Abstract

The purpose of this paper is to report a rare form of Brooke-Spiegler syndrome with collision tumor (syringocystadenoma papilliferum with cylindromas). The Brooke-Spiegler syndrome is a genodermatosis of dominant autosomal inheritance. It is characterized by the development of multiple benign tumors of the skin appendages from the change in the sebaceous-apocrine follicular unit. The appendages tumors most commonly found in the syndrome are cylindromas, trichoepitheliomas and eccrine espiroadenomas. We report the case of a 59-year-old male patient, who had had since puberty an onset of papular and nodular lesions in the head, neck and back. This paper highlights the presence of tumor in the collision scalp.

Resumen

El objeto de este artículo es informar sobre una forma poco frecuente del síndrome de Brooke-Spiegler con tumor de colisión (syringocystadenoma papilliferum con cilindromas). El síndrome de Brooke-Spiegler es una genodermatosis de herencia autosómica dominante. Se caracteriza por el desarrollo de múltiples tumores benignos de los apéndices de la piel debido al cambio en la unidad folicular sebáceo-apocrina. Los tumores de los apéndices que se encuentran con mayor frecuencia en el síndrome son los cilindromas, los tricoepiteliomas y los espiroadenomas ecrinos. Reportamos el caso de un paciente varón de 59 años, que desde la pubertad presentaba lesiones papulares y nodulares en cabeza, cuello y espalda. Este artículo destaca la presencia de un tumor en el cuero cabelludo en colisión.

INTRODUCTION

Brooke-Spiegler syndrome (BSS) is an autosomal dominant disease with variable expression and penetration.[1,2] The manifestations of the disease, even being a genodermatosis one, usually occur between the second and the third life decades, mainly within the female sex and it has a wide range of tumors with eccrine, apocrine, follicular and sebaceous differentiation. Occasionally, one same tumor has different associated cell groups, such as the spiroadenocilindroma.[3] It is presented here a case of a patient with exuberant Brooke-Spiegler syndrome associated with a collision tumor in the scalp.

CASE PRESENTATION

A male patient, white skin colored, 59 years old, with multiple papular and nodular lesions, asymptomatic, mainly at his back, head and neck, having a progressive growth from puberty. Six years ago, it refers to the appearance of new nodular lesions on the face and scalp. The patient was unaware of a similar family history background. At the examination, there were multiple papules and reddish, firm pinkish nodules of varying sizes (within 1-5 cm), with a smooth and shiny surface, arranged in an isolated form on the scalp and on the face *(Figure 1)* and confluent in the retro-auricular area *(Figure 2)*. He also presented pink papular lesions on the face, with telangiectasia on the surface *(Figure 2)*, and normocromic papules on his back *(Figure 3)*. In the nasal dorsum he presented a pearlescent papule with arboriform telangiectasia and ovoid nodules to dermoscopy. It was carried out incisional and other excisional biopsies, being the histopathological one compatible with (1) tricoepithelioma on the face; (2) cylinder in the retro-auricular region and back and (3) collision tumor (papilliferous syringocystadenoma with cylinder) on the scalp *(Figures 4 to 6)*. Due to the clinical findings, histopathological ones and to onset at the puberty period, we have noticed that it was about the Brooke-Spiegler syndrome, which was associated to the collision tumor. Due to the psychosocial impairment made by the unsightly

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aspect of the lesions, surgical excision of the major lesions and of the basal cell carcinoma suspected lesion in the nasal dorsum are scheduled.

DISCUSSION

Brooke-Spiegler syndrome is characterized by mutations within the CYLD tumor suppressor gene, also named the cylindromatosis gene, located within the chromosome.[1,4] This gene has the function of regulating the correct proliferation of the cutaneous appendages through the synthesis of the CYLD protein, an enzyme that negatively regulates the transcription factor NF-kB, which is an inducer of adnexal proliferation.[3,5] The gene alterations cause defects in the differentiation of the apocrine pilosebaceous unit, originating different adnexal tumors.[3] Normally cranberry, trichoepitheliomas and eccrine spiradenoma are found, originating from the basal layer of the epidermis and hair follicles, located mainly in the head and in the neck zones.[6,7] The diagnosis of this syndrome does not require the presence of the three types of tumors, being only necessary the presence of two of them. Associations of this syndrome

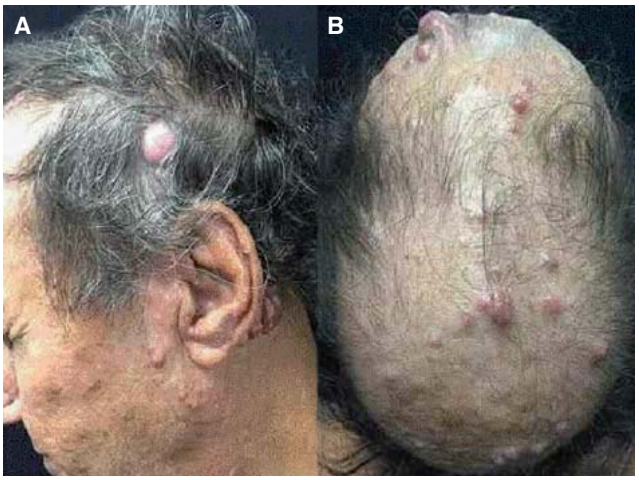

Figure 1. Rosy papules and nodules, smooth and shiny surface, having varying sizes, on the scalp and face.

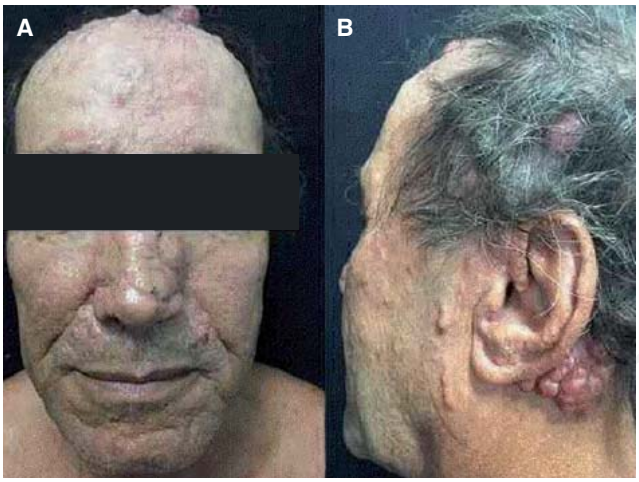

*Figure 2. **A:** On the face, there are slightly pinkish papules with telangiectasias on the surface. **B:** Pink tumor lesion in the retro-auricular region, having a measuring approximately 5 x 6 cm.*

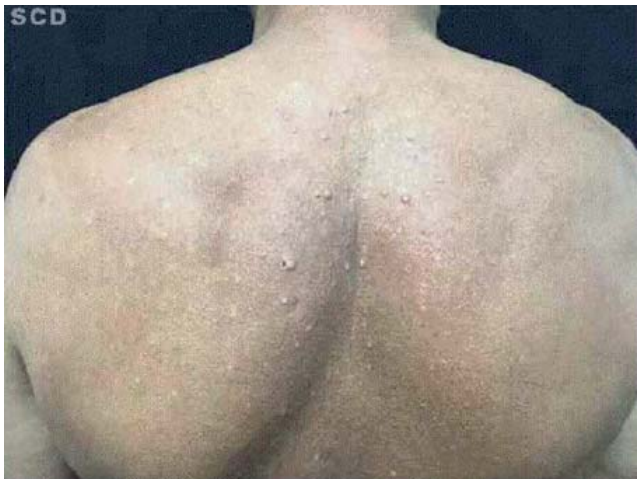

Figure 3. Normachromic papular lesions on the back.

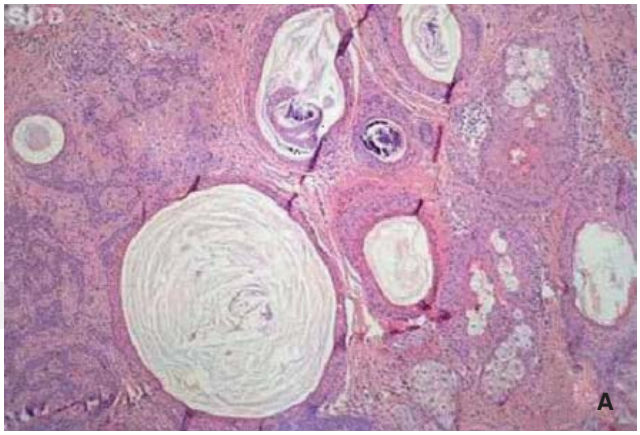

*Figure 4. **A:** Basal cell masses, with peripheral palisade, horny cysts and dense stroma.*

with basal cell carcinomas were identified, sebaceous nevus, milium, adenoma and carcinoma of parotid glands, salivary and submaxillary glands, xeroderma pigmentosum, hypo and hyperchromias, polycystic disease in the lungs, kidneys, breast and multiple fibroids.[8] There are reports of malignant transformation of dermal cylindroma, and lymph node, thyroid, liver, lung and bone metastases may occur.[9] The association of contiguous tumors is not uncommon, although clinical diagnosis is difficult within isolation. The most common association is the basal cell carcinoma and nevus one.[10] In the case report aforementioned, the presence of an atypical collision tumor, papillary syringocystadenoma with cylinder, was noted, with few reports in the scientific literature. Although some of these associations may occur through the involvement of related cell types, most of them occur at random. The treatment of SBS should be directed towards the possibility of malignant transformation and the progressive nature of the disease. Generally speaking, due to the refractoriness of tumors to conventional treatments, surgical excision is the chosen method whenever it is possible. The clinical monitoring of patients, due to the possibility of malignant transformation and association with other neoplasms. Genetic counseling is also necessary.[3]

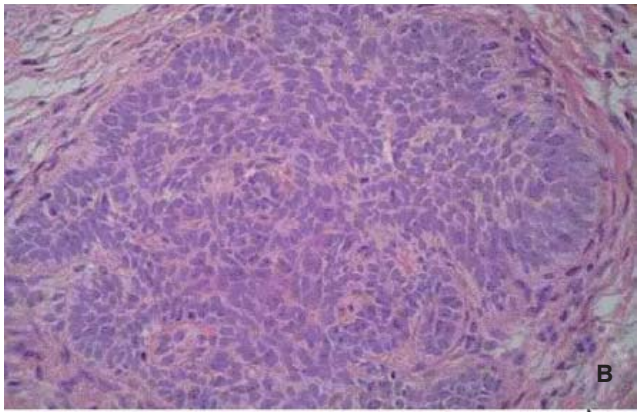

Figure 4. ***B:*** *Detail of the basaloid mass of follicular origin and cribriform aspect.*

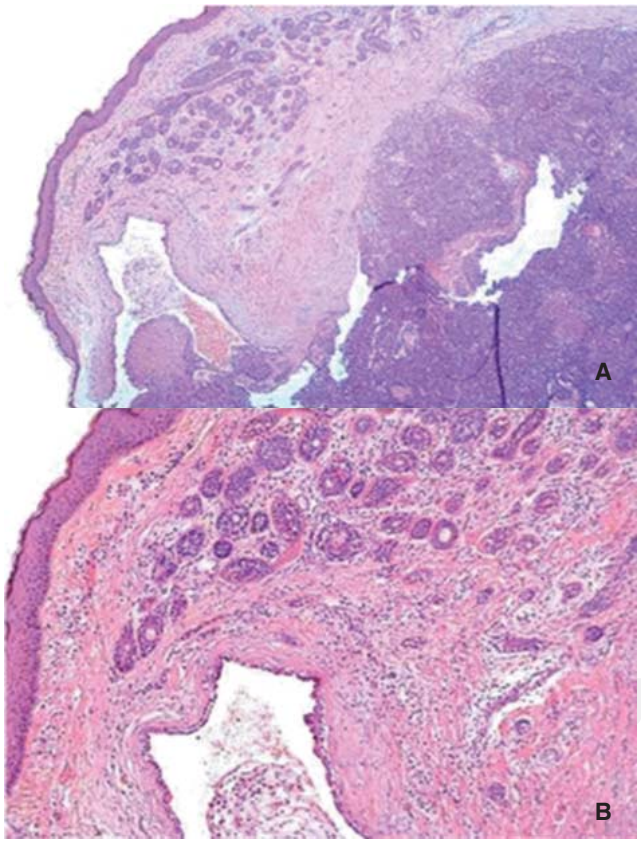

Figure 5. HE, 40 and 100x: Papillary projection area with columnar cells and decapitation secretion.

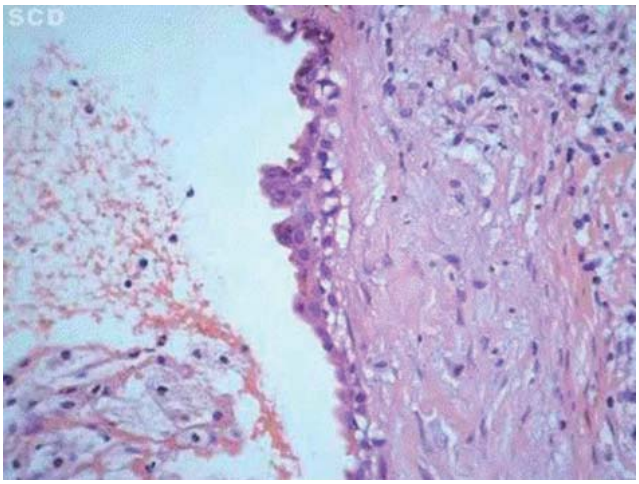

Figure 6. HE, 40 and 100x: Papillary projection area with columnar cells and decapitation secretion.

CONCLUSIONS

It is presented here a rare case of a patient with exuberant Brooke-Spiegler syndrome associated with a collision tumor in the scalp.

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