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Case Reports

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Giant eccrine spiradenoma associated with Brooke-Splieger syndrome

Espiradenoma écrino gigante associado à síndrome de Brooke-Splieger

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ABSTRACT

Brooke-Spiegler syndrome is a rare autosomal dominant genetic disease with predisposition to many adnexal tumors, including trichoepithelioma, cylindroma and spiroadenoma. Tumors appear in the second decade of life, progressively increase with age, and their prevalence is higher in women. It is caused by a mutation in the CYLD gene, localized in the chromosome 16q12-q13. We report a exuberant case of giant eccrine spiradenoma associated to this syndrome.

Keywords: Dermatologic Surgical Procedures; Facial Neoplasms; Neoplasms, Adnexal and Skin Appendage

RESUMO

A síndrome de Brooke-Spiegler é doença genética autossômica dominante rara, com predisposição a diversos tumores anexiais, dentre eles tricoepitelioma, cilindroma e espiradenoma. Os tumores surgem na segunda década de vida, aumentam progressivamente com a idade e sua prevalência é maior em mulheres. É causada por mutação no gene CYLD, localizado no cromossomo 16q12-q13. Relatamos caso exuberante de espiradenoma écrino gigante associado a essa síndrome.

Palavras-Chave: *Neoplasias Cutâneas; Neoplasias de Anexos e de Apêndices Cutâneos; Procedimentos Médicos e Cirúrgicos de Sangue*

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INTRODUCTION

The Brooke-Spiegler syndrome is a rare hereditary autosomal dominant inheritance caused by a mutation in the CYLD gene, which is located on chromosome 16q12-q13.[1]

It has the clinical appearance of cylinder type multiple adnexal tumors, trichoepithelioma and spiradenoma.[2]

The authors of the present report describe a rare case of Brooke-Spiegler syndrome associated with giant eccrine spiradenoma in the forehead.

CASE REPORT

A previously healthy 55-year-old male patient reported an eight-year the history of a progressive, slow-growing forehead mass. In addition to the aesthetic impairment, the patient complained of decreased visual field due to the lesion. At the dermatological examination, it was possible to observe a well delimited, fibroelastic, pedunculated, normochromic-reddish

and elevated frontal tumoral mass measuring 9.0x4.0x7.0cm (Figures 1 and 2). The patient had multiple small normochromic nodules on the face and scalp.

There were similar cases of multiple face nodulations in the patient's family. The lesion was excised (Figure 3) and excess skin was removed for primary closure.

Figure 4 shows the immediate postoperative period, while Figure 5 shows the 10th day after surgery. The patient was extremely satisfied with the surgical outcome.

The anatomopathological examination evidenced multiple foci of juxtaposed basaloid blocks and other clear cells located in the superficial and deep reticular dermis, forming nodules with precise and lobed borders, compressing the adjacent subcutaneous tissue and forming a pseudo-capsule. There are areas of necrosis with formation of cystic structures filled by fibrinoid and hematic material. Blind bottom ducts are also observed

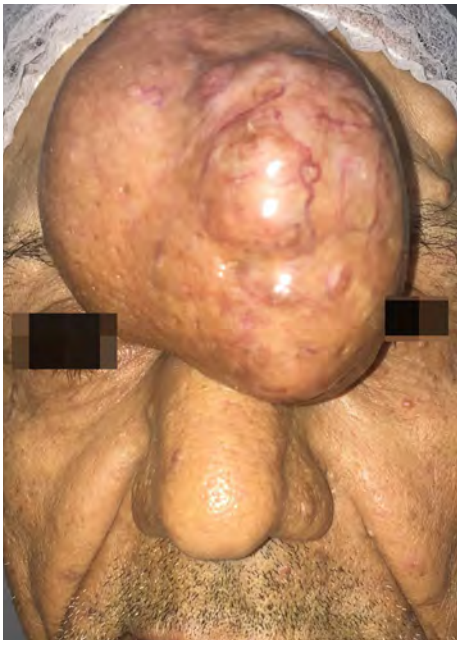

Figure 1: Normochromic, well-delimited tumor in the forehead, with vessels on the surface. Notice the minor nodulation in the left hand side of the upper forehead

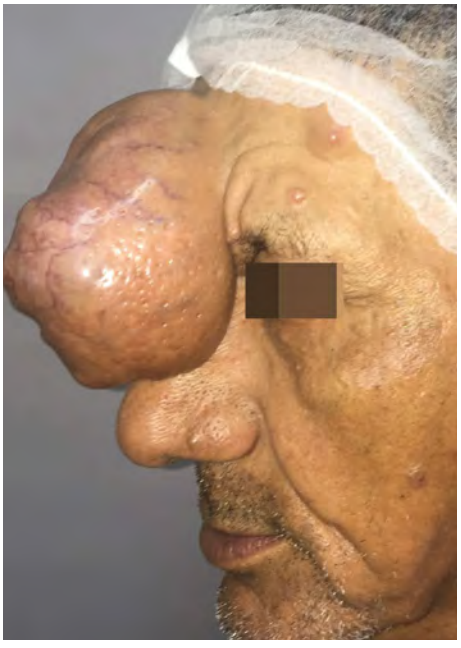

Figure 2: Lateral view of pedunculated tumor in the forehead

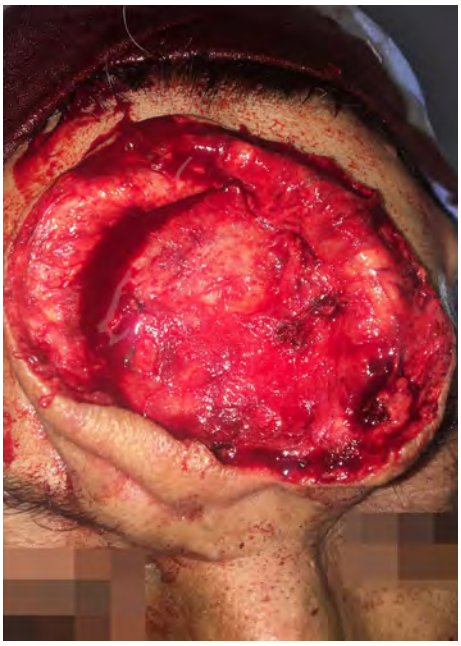

Figure 3: Image demonstrating the intraoperative period

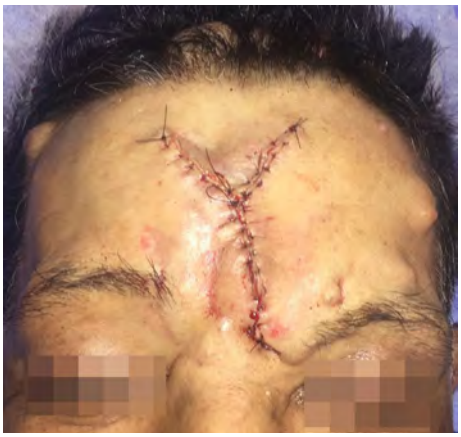

Figure 4: Immediate postoperative period

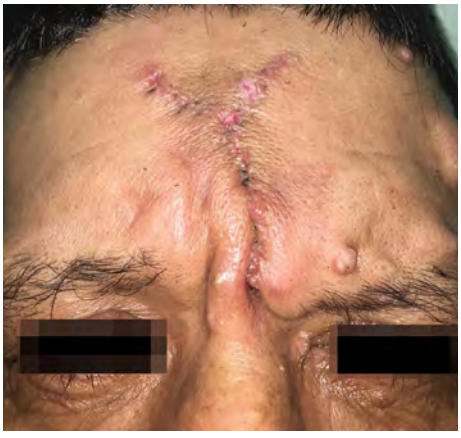

Figure 5: Tenth postoperative day

(Figures 6 and 7). Immunohistochemistry was performed for BEREP4, which came out negative, and for Ki-67, which was positive in 15% of the neoplastic cells. These findings suggest a diagnosis of eccrine spiradenoma.

DISCUSSION

The Brooke-Spiegler syndrome is a rare autosomal dominant disease characterized by the development of multiple adnexal neoplasms, including cylindroma, spiradenoma, and trichoepithelioma. [3] It was first reported in 1842 by Ancell. [2] It has higher prevalence in women.[4,5] CYLD, the gene is implicated in the pathogenesis of the disease, is a tumor suppressor gene located on the chromosome 16q12-q13. In addition to the skin, morphologically similar neoplasms may arise in the salivary glands and breasts, however this is extremely rare.[1]

Patients with Brooke-Spiegler syndrome have multiple tumors located mainly in the head and neck region. Most nodules measure 0.5 to 3.0 cm, however larger lesions can also be found[1], as in the present case.

Most of the tumors microscopically correspond to spiradenomas, cylindromas or trichoepitheliomas. They are histologically identical to sporadic cases; however, in cases of this syndrome, it is more common to find variants of multifocality of tumor types in the same lesion.

Cylindromas occur as numerous papules, nodules or tumors distributed on the scalp and sometimes on the face and trunk.[6] A classic presentation of multiple confluent lesions on the scalp is called a "turban tumor".[7] They are histologically characterized by a well-circumscribed lesion composed of tumor islands and basaloid cell cords organized in a "puzzle" pattern.[8] Malignant transformation of cylindroma is rare.

In addition to scalp lesions, patients with the classic Brooke-Spiegler syndrome phenotype have small, normochromic discrete and / or confluent papules, of 0.2 to 1.0 cm in size, located in the nasolabial folds, histologically corresponding to trichoepitheliomas. These are aggregates of basaloid cells with peripheral palisade formation, relatively monomorphic in the dermis, surrounded by fibrous stroma. Retraction artifacts and mucinous stroma are absent in this tumor.[8]

In the present case, the patient had a giant eccrine spiradenoma. This tumor is histologically characterized by cell lobes – often encapsulated and circumscribed by basaloid cells – filling the dermis. Small ductal lumens can be seen in the centers of the lobes. There is no cellular pleomorphism, and mitotic activity is sparse or absent. There can be lymphocytic infiltration into the tumor.[8]

The tumors appear mainly in the second decade of life and their quantity increases progressively with age. They grow slowly and progressively. Rapid growth associated with ulceration and bleeding should raise the suspicion of malignant transformation.[4,5] Malignant tumors arise in association with pre-existing benign cutaneous neoplasms in about 5 to 10% of patients.[1,6]

The Brooke-Spiegler syndrome, multiple familial trichoepithelioma, and familial cylindromatosis share overlapping clinical findings. While patients with Brooke-Spiegler syndrome are predisposed to multiple adnexal tumors, patients with familial cylindromatosis have only cylindromas, [2] and those with multiple familial trichoepitheliomas, only have trichoepitheliomas.[1]

The different treatment methods suggested for adnexal tumors include excision, dermabrasion, cryotherapy and CO_2 laser. For eccrine spiradenoma and cylindroma, surgery is the

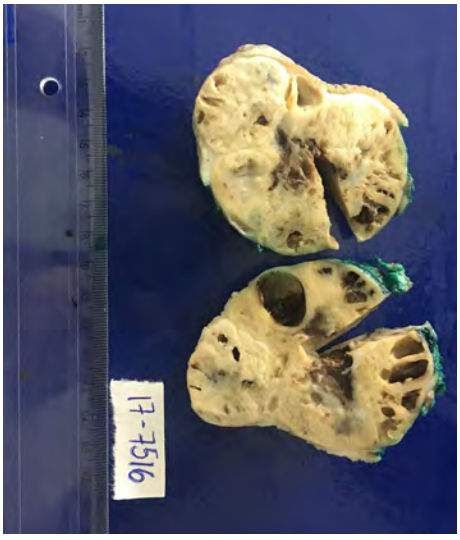

Figure 6: Surgical specimen showing a well-delimited tumor composed of cystic areas in its interior

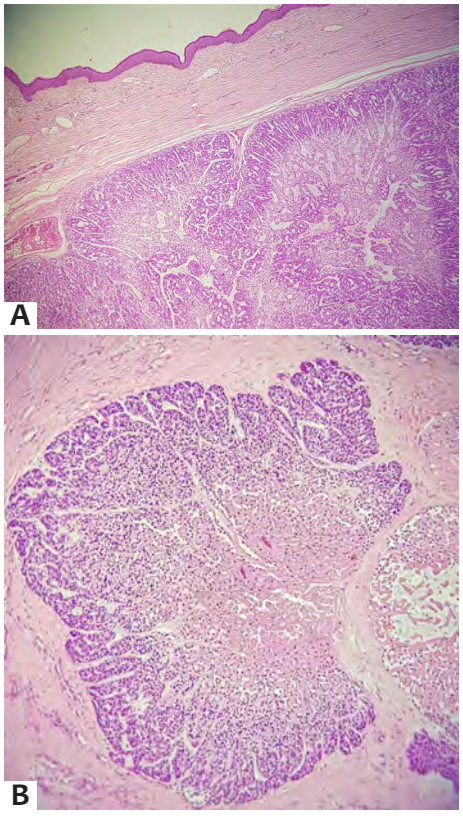

Figure 7: A - Anatomopathological examination evidencing focus of lobulated basaloid block compressing adjacent subcutaneous tissue and forming a pseudo-capsule. **B -** Pathologic examination evidencing focus of lobed basaloid block

treatment of choice. [3] In the case described in the present paper, the authors made a choice for surgical treatment due to the extension of the lesion and aesthetic compromise. Surgical treatment of eccrine spiradenoma is curative.

CONCLUSION

The Brooke-Spiegler syndrome is a rare genetic disease predisposing to adnexal cutaneous tumors. The authors of the present report describe an exuberant case of giant eccrine spiradenoma associated with this syndrome, with excellent surgical outcome. ●

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