Giant eccrine spiradenoma associated with Brooke-Spiegler syndrome

Authors: Taiane Medeiros Terra¹
Flavia Tandaya Grandi Miranda¹
Luiz Fernando Froes Fleury Junior¹

¹ Department of Dermatology, Universidade Federal de Goiás - Goiânia (GO), Brazil.

ABSTRACT

Brooke-Spiegler syndrome is a rare autosomal dominant genetic disease with predisposition to many adnexal tumors, including trichoepithelioma, cylindroma and spiradenoma. 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

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
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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 basaloïd 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 fibri-noid 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. It was first reported in 1842 by Ancell. It has higher prevalence in women. 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.

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, 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. A classic presentation of multiple confluent lesions on the scalp is called a “turban tumor”. They are histologically characterized by a well-circumscribed lesion composed of tumor islands and basaloid cell cords organized in a “puzzle” pattern. Malignant transformation of cylindroma is rare.

In addition to scalp lesions, patients with the classic Brooke-Spiegler syndrome phenotype have small, normochromatic 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.

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.

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. Malignant tumors arise in association with pre-existing benign cutaneous neoplasms in about 5 to 10% of patients.

The Brooke–Spiegl er 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, and those with multiple familial trichoepitheliomas, only have trichoepitheliomas.

The different treatment methods suggested for adnexal tumors include excision, dermabrasion, cryotherapy and CO2 laser. For eccrine spiradenoma and cylindroma, surgery is the preferred method due to their slow growth and potential for malignant transformation.
REFERENCES


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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DECLARATION OF PARTICIPATION:

Taiane Medeiros Terra | ORCID 0000-0002-6479-8686
Approval of the final version of the manuscript; study design and planning; preparation and drafting of the manuscript; intellectual participation in propaedeutic and / or therapeutic treatment of cases studied; critical review of the literature.

Flavia Tandaya Grandi Miranda | ORCID 0000-0002-4323-2499
Resident Doctor in Dermatology, Dermatology Service of the Universidade Federal de Goiás, Goiânia (GO), Brazil

Luiz Fernando Froes Fleury Junior | ORCID 0000-0002-1202-6211
Dermatologic Surgery, Dermatology Service of the Hospital das Clínicas of the Universidade Federal de Goiás