Case Report

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Brooke spiegler syndrome-a case report

Abstract

Brooke Spiegler syndrome is rare autosomal dominant hereditary disease with variable penetration. It is more common in females, usually in the second and third decades of life. The syndrome manifests with the appearance of three types of tumors: multiple cylindromas, trichoepitheliomas and spiradenomas. We presented a female patient, 54 years of age. The first changes occurred at the age of 22, first on the forehead, and then on the scalp. She has a positive family history for this condition, obtained by anamnesis (mother, two sisters, brother and daughter).

We presented a dermatologic status before and after surgical intervention. Pathohistological finding of the removed tumors: cylindroma. After surgical intervention, the patient's quality of life has improved. New surgical procedures are planned.

Keywords: brooke-spiegler syndrome, cylindroma, trichoepithelioma, spiradenoma

Volume 2 Issue 1 - 2018

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Received: November 24, 2017 | **Published:** January 23, 2018

Introduction

Brooke Spiegler syndrome (BSS) is a rare autosomal dominant hereditary disease with variable penetration.[1,2] It is more common in females,[3] usually in second and third decades of life.[4] Brooke Spiegler syndrome is manifested by the appearance of three types of tumors. Cylindroma's are different number and size, from few to multitude, covering the entire scalp, usually without any symptoms. Typical trichoepithelioma in the form of yellowish papules and small tumorous nodules are usually on the face, and they cause psychosocial problems in patients due to location.[5] Spiradenoma, in the form of individual tumorous formation due to the location is most frequently localized in the upper part of the trunk, upper extremities, and rarely on the chest.

Case report

LM 54 year's old female. She has a son and a daughter. She has a positive family history obtained by anamnesis (mother, two sisters, brother and daughter) (Figure 1). First changes occurred when she was 22, first on the forehead, and then on the scalp.

Status dermatological (before the surgery): On capillitium multiple well-circumscribed, pink, nodular cylindroma, of 1 to 7cm in diameter, with arborizing blood vessels on the surface, especially dense at top of the head, but also on the forehead and other areas, slightly rear at the back of the head (Figure 2).They can be found before and retroauriculary in the outer ear canals. They are tumefacts in skin colour or pink, with smooth shiny surface, occasionally with telangiectasias (Figure 3). On the forehead, skin is uneven with unevenly elevated papules of yellowish colour to the skin colour (Figure 4).On the back individual tumorous formations, with characteristic blue/black appearance, of 1cm in diameter but underneath, tumorous nodules up to the size of wall nut (typical for spiradenoma). Similar changes in lower number are found in the front part of the trunk (Figure 5).

Surgical treatment consisted of total excision on the scalp and application of skin grafts for restoracion. Post surgical status: Skin of capillitium and half of forehead after the surgery and skin grafts shiny and smooth, without hair, with occasionally lowered sensitivity (Figure 6).

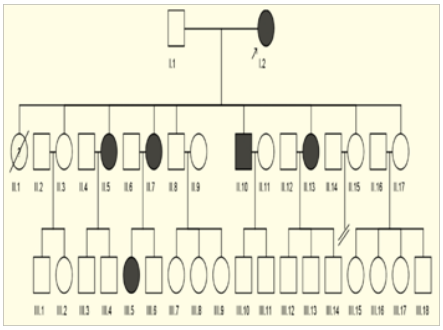

Figure 1 Genetic pedigree.

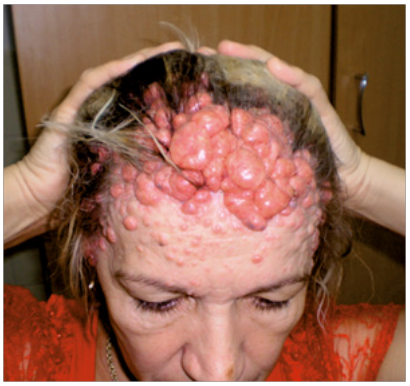

Figure 2 On capillitium multiple tumor formations.

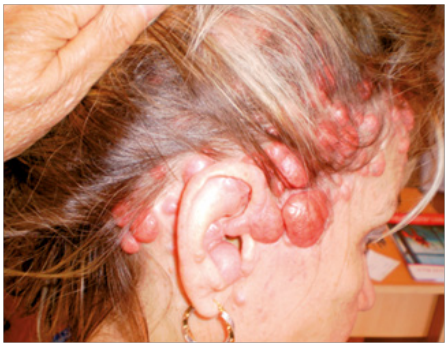

Figure 3 Tumor formations before and retroauriculary.

Figure 4 On the forehead skin uneven with unevenly elevated papules.

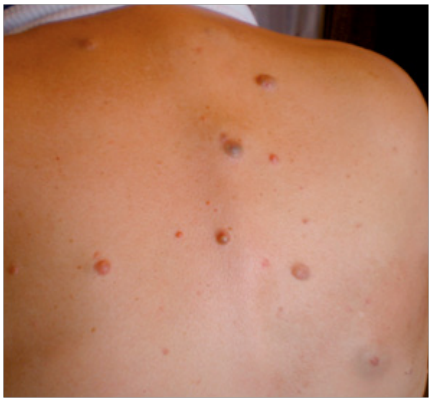

Figure 5 On the back individual tumors formations of skin colour.

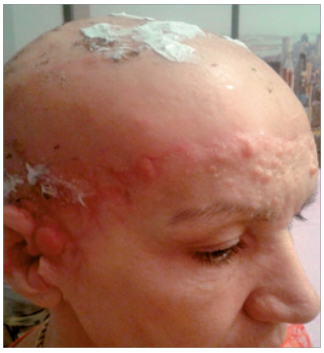

Figure 6 Skin of capillitium and a half of forehead shiny and smooth, without hair.

It is planned to remove other tumors as well. Pathohistological finding on the removed tumors: The typical appearance of a cylindroma at low power (10x), consisting of well-defined nests of basaloid cells separated by an eosinophilic basement membrane. Biopsy of face and back tumors was not performed. However, it was performed in the brother and sister and it was proved that they were trichoepitheliomas and spiradenomas.

Discussion and conclusion

Genetic studies determined that BBS is caused by heterozygote mutation of cylindromatose gene. CYLD[6–8] located on the chromosome 16q12-q13.[9] Today, it is believed that BSS, as earlier described individual diseases Ancell-Spigler syndrome and Brooke Fordyce trichoepithelioma are the consequence of defects in putative stem cells regulation of foliculo sebaceous apocrine unit, what enables development of various skin tumors.[10,11]

CYLD gene has tumor suppressing function in the development of different cancers,[7] so that persons with tumor suppressor gene CYLD mutations are at high risk of development of these tumors. BSS is not curable. Besides the application of different medicines and methods, surgical therapy is most frequently used in order to improve cosmetic aspects as well as to prevent and minimize complications.[12] Brooke Spiegler syndrome is a rare hereditary disease. In our patient, surgical intervention gave satisfactory therapeutic results, what contributed to the improvement of her quality of life.

Acknowledgements

None.

Conflicts of interest

The authors declared that there are no conflicts of interest.

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