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Brooke Spiegler Syndrome - A Case Report

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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. New surgical procedures are planned.

Keywords: Brooke Spiegler syndrome, Cylindroma, Trichoepithelioma, Spiradenoma

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. Brooke Spiegler syndrome is manifested by the appearance of three types of tumors. Cylindroma, of different number and size, from few to multitude, covering the entire scalp, usually without any symptoms, typical trichoepithelioma and spiradenoma [4].

CASE REPORT

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

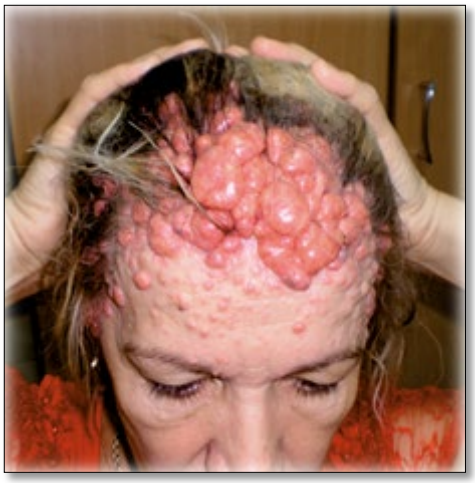

Figure 1. Status dermatologicus.

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Status dermatologicus (before the surgery): On capillitium multiple well-circumscribed, pink, nodular cylindroma, with arborizing blood vessels on the surface of 1 to 7 cm in diameter especially dense at top of the head, but also on the forehead.

They are tumefacats in skin color or pink, with smooth shiny surface, occasionally with telangiectasias. On the forehead, skin is uneven with unevenly elevated papulas of yellowish color to the skin color. On the back individual tumorous formations, with characteristic blue/black appearance, of 1 cm in diameter but underneath, tumorous nodules up to the size of walnut (typical for spiradenoma). Similar changes in lower number are found in the front part of the trunk surgical treatment consisted of total excision on the scalp and application of skin grafts for restoration. Post-surgical status: Skin of capillicium and half of forehead after the surgery and skin grafts shiny and smooth, without hair, with occasionally lowered sensitivity **(Figure 2)**.

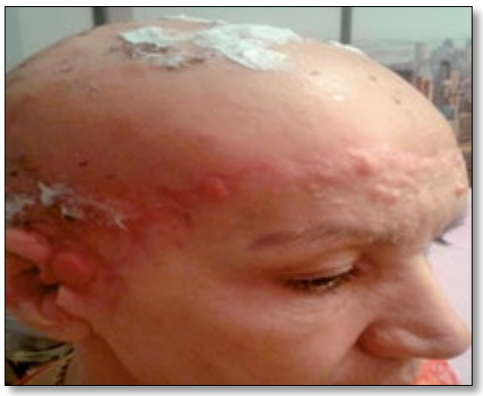

Figure 2. Post-surgical status.

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

DISCUSSION AND CONCLUSION

Genetic studies determined that BBS is caused by heterozygote mutation of cylindromatose gene. CYLD [5] located on the chromosome 16q12-q13 [6]. 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 minimise complications [7]. 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.

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