 DOI: 10.18231/2348-7240.2017.0007

Brooke-spiegler syndrome in a 35 years old female: Case report with review of literature

Anand Dutta[1,*], Nirbhaibie Singh[2], Sahil Mashal[3], Devika[4], Gaurav Sharma[5], Parneet[6]

[1,2,3,4,5,6]Junior Resident, Dept. of Surgery, Govt. Medical College, Amritsar, Punjab

***Corresponding Author:**
Email: dranandduttta@gmail.com

Abstract

Brooke-Spiegler syndrome (BSS) was reported for the first time in 1842 by Ancell.(1) It is an autosomal dominantly inherited syndrome, characterised by multiple skin appendageal tumors especially trichoepitheliomas and cylindromas and occasionally spiradenomas which usually appears in the second or third decade of life.(2) With just 60 cases of Brooke-Spiegler syndrome reported in the literature so far across the globe, the entity is considered very rare. Herein, we are reporting a case of Brooke-Spiegler Syndrome in 35 years old female.

Keywords: Brook-Spiegler syndrome; Cylindroma; Trichoepithelioma

Case Report

A 35 years old female presented in surgery department with complaint of swelling of about 0.5cm * 1cm in right parotid region since 6 months. The patient also had multiple cutaneous lesions on her face. These lesions first appeared when she was 14 years old, primarily on her face and had gradually increased in size and number over the years. The patient's sister have similar complaints and her son had similar facial lesions as well. On examination, a group of round-to-oval skin-colored papules with a smooth pearly surface measuring 2 to 6 mm in diameter was seen on the mid-face, particularly in the nasolabial folds and the upper lip (Fig. 1, 2). Her general condition was good. She had no known underlying disease and no previous history of taking any kind of medications. The swelling was excised under local anaesthesia and the histopathology revealed a dermal lesion with well-defined borders composed of islands of basaloid cells arranged in a jigsaw puzzle-like pattern and separated by hyaline basement membrane material. Two populations of basaloid cells (with a large nucleus centrally and smaller nuclei in a palisading pattern on the periphery) were observed, suggestive of cylindroma (Fig. 3, 4).

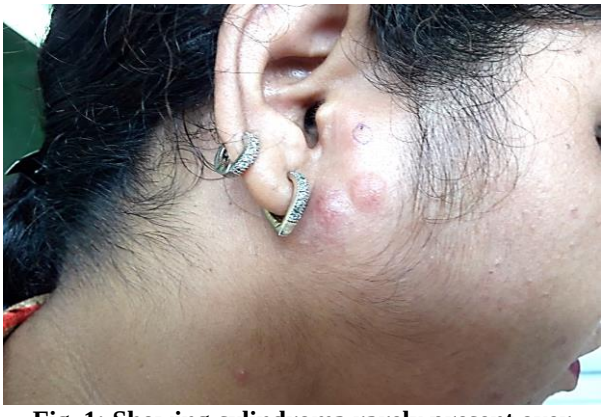

Fig. 1: Showing cylindroma rarely present over parotid region

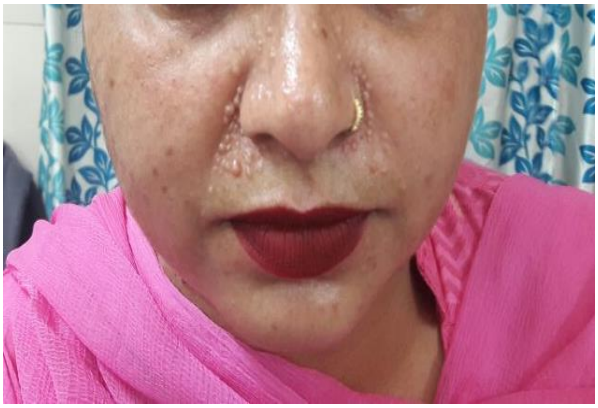

Fig. 2: Showing multiple trichoepitheliomas around nasal area

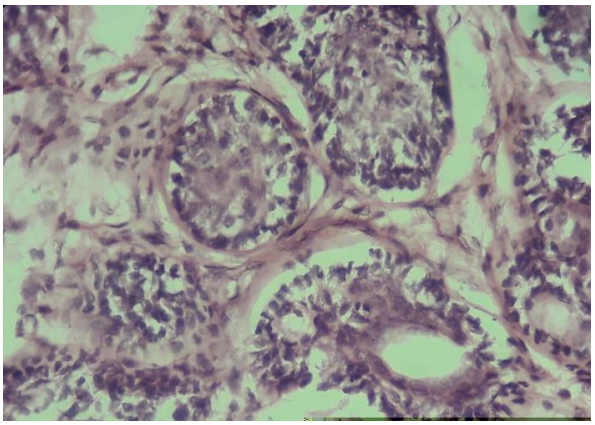

Fig. 3: Showing a dermal lesion with well-defined borders composed of islands of basaloid cells

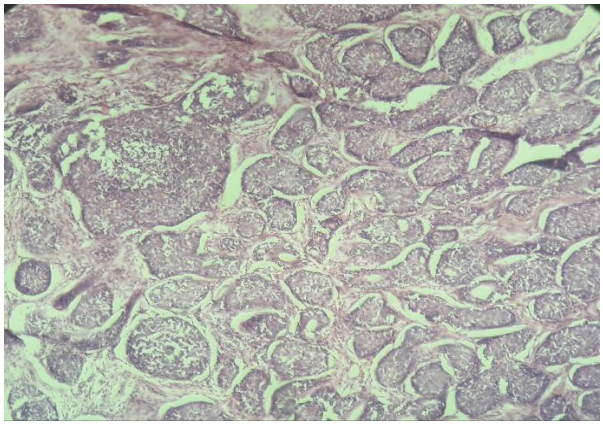

Fig. 4: Showing a large nucleus centrally and smaller nuclei in a palisading pattern on the periphery

Discussion

BSS has an autosomal dominant mode of inheritance and tends to form numerous adnexal tumors, in particular trichoepithelioma, cylindroma, and occasionally spiradenoma, especially in the second or third decade of life.[1] To date, the cell-type specific origin of BSS is a topic of ongoing debate. For cylindroma and spiradenoma, both the apocrine and eccrine origins have been suggested, but the origin of follicular trichoepithelioma is not yet confirmed.[2] Some individuals may present with isolated trichoepithelioma or cylindroma, whereas others can have a combination of them in one tumor, as described by Honag Ly *et al,* which suggests a common embryonic relationship between the follicles and the apocrine glands.[3] Thus BSS represents a genetical disorder which affects the regulation of the folliculo-sebaceous apocrine unit.

Further evidence for this hypothesis is provided by reports of other follicular tumors, such as BCC, follicular cyst, and nevus sebaceous, associated with this syndrome.[4] These findings indicate that a mutation in the genes that regulate the stem cell proliferation and differentiation can cause a defect in the differentiation of the folliculo-sebaceous apocrine unit, which gives rise to different combinations of adnexal skin tumors.[5] Genetical studies have demonstrated that mutations in the CYLD 1 gene are responsible in this disease.[6] The penetrance of the gene has been estimated to be between 60% and 100%. The disease also demonstrated variable clinical and histopathological features among the affected members of a single family, in which older members tend to have larger lesions that are also greater in number as compared to the younger members.[7] This condition was obvious in our patient's family, where her son had numerous trichoepithelioma lesions on his face and her sister had also similar complaints. Another fact which should be importantly taken into consideration in this case is that such patients are at risk for developing BCC from trichoepithelioma, and they should be followed up and clearly informed to visit the clinic if the lesions increase in number or become ulcerated.[8] The different suggested modes of treatment for the adnexal tumors in BSS patients include excision of the tumor, dermabrasion, electrodessication, cryotherapy, and radiotherapy using argon and CO2 lasers.

References

1. Bandmann HJ, Hamburger D, Romiti N. Brooke-Spigler phakomatose. Hantarzt. 1965;16:450-3.
2. Weyers W, Nilles M, Eckert F, Schill WB. Spiradenoma in Brooke-Spigler syndrome. Am J Dermatopathol. 1993;15:156-61.
3. Ly H, Black MM, Robson A. Case of the Brooke-Spiegler syndrome. Australian J Dermatol. 2004;45:220-2.
4. Ferrandiz C, Campo E, Bauman E. Dermal cylindromas (turban tumor)and eccrine spiradenomas in a patient with membranous basal cell adenoma of the parotid gland. J Cutan Pathol. 1985;12:72-9.
5. Fenske C, Banerjee P, Holden C, Carter N. Brooke-Spiegler syndrome locus assigned to 16q12-q13. J Invest Dermatol. 2000;114:1057-8.
6. Young AL, Kellermayer R, Szigeti R, Tıszαs A, Azmi S, Celebi JT. CYLD mutations underlie Brooke-Spiegler, familial cylindromatosis and multiple familial trichoepithelioma syndromes: Short report. Clin Genet. 2006;70:246-9.
7. Saunders H, Tucker P, Saurine T, Watkins F. Pedigree of multiple benign adnexal tumors of Brooke-Speigler type. Australas J Dermatol. 2003;44:144-8.
8. Rallan D, Harland CC. Brooke-Spiegler syndrome: Treatment with non ablative laser. Clin Exp Dermatol. 2005;30:355-7.