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CASE REPORT

A 54-year-old man reported a painless retroauricular lesion, with progressive growth over 40 years, and lesions on the face and scalp for two years. He had hypertension without other comorbidities, and reported past surgery on the nasal crease with a diagnosis of trichoepithelioma. There were no children or family history.

At examination he presented: frontal-parietal alopecia; (1) a 3.5 x 3 cm multilobular violaceous nodule of elastic consistency in the left retroauricular region; (2) multiple erythematous papules and nodules on the scalp, with elastic consistency and telangiectasias (turban aspect); (3) normochromic papules on the upper and lower eyelids; (4) a 1 x 1 cm nodule with a pearlaceous border and telangiectasias in the left nasal region; (5) a 2 x 2 mm papule with a pearlaceous border and telangiectasias in the left nasogenian groove; (6) multiple normochromic millimetre size papules on the back (Figure 1).

Incision biopsies performed on these lesions revealed: cylindroma (1, 2, and 6); trichoepithelioma (3 and 5), and trichoblastoma (4) (Figures 2 e 3).

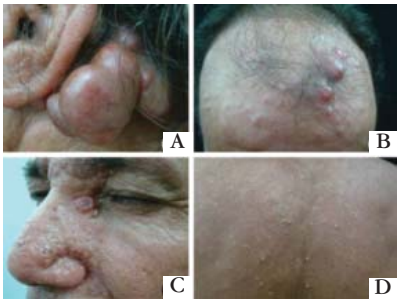

FIGURE 1: A - Multilobular violaceous nodule in the left retroauricular region; **B -** erythematous papules and nodules on the scalp, with telangiectasias (turban aspect); **C -** normochromic papules on eyelids; nodule with a pearlaceous border and telangiectasias in the left nasal region; normochromic papule in the left nasogenian groove; **D -** normochromic papules on the back

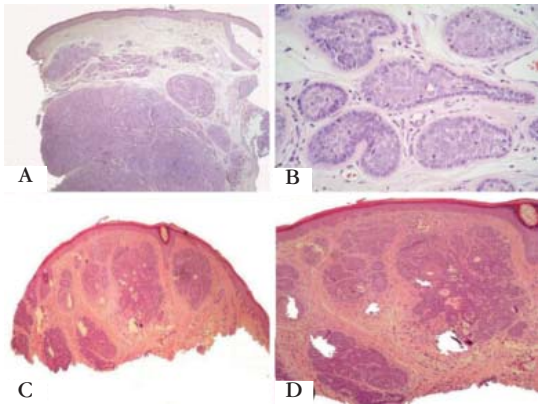

FIGURE 2A: Cylindroma (HE 40x): base cell tumour arranged in islands in the dermis, some with sudoriparous ducts; **2B:** Cylindroma (HE 400x): hyalinised cylinders surrounding each tumour island. **2C:** Trichoepithelioma (HE 40x): islands of well demarcated basaloid tumour cells and some horn cysts; **2D:** Trichoepithelioma (HE 100x): peripheral distribution nucleus palisades, without retraction artefact or loose stroma

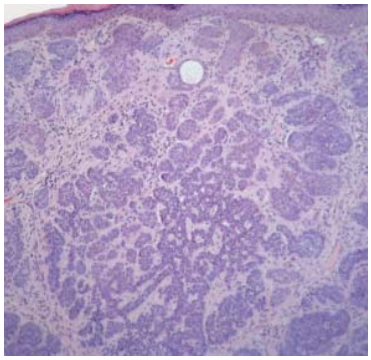

FIGURE 3: Trichoblastoma (HE 100x): Dermal proliferation of basaloid cells without atypical fibrotic stroma

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DISCUSSION

Brooke-Spiegler Syndrome (BSS) is an autosomal dominant disorder with variable expression and penetration which appears in the second and third decades of life and is more prevalent in women. They are characterised by a genetic predisposition to develop adnexal neoplasias on the head and neck. In practice, patients are seen to develop neoplasias with eccrine, apocrine, follicular, and sebaceous differentiation, making it possible for the same neoplasia to present different groups of cells (e.g., spiradenoma cylindroma).[1]

BSS results from mutations or loss of heterozygosity in the cylindromatosis gene (CYLD) located in chromosome 16q12-q1.[2] The cylindromatosis gene is a tumour suppressor, and its product suppresses the tumour necrosis factor-α (TNF-α) route. Activation of this route increases expression of nuclear factor -κβ (NF-κβ), a transcription factor which regulated the number of anti-apoptotic genes involved in the proliferation of skin adnexal neoplasms. Mutations in the CYLD gene result in increased NF-κβ expression leading to apoptosis resistance and the appearance of pilosebaceous apocrine unit tumours: cylindromas, trichoepitheliomas, and spiroadenomas.

This syndrome has associations with other tumours such as: basal cell carcinomas, sebaceous nevus, milium, parotid adenoma and carcinoma, xeroderma pigmentosum, hypo and hyperchromia, poly cystitis, and fibromas.[3]

Cylindromas present in two forms: as a difficult to diagnose solitary lesion most frequented located on the head; or as multiple different sized erythematous nodules on the scalp which can flow together resembling a turban. There are reports of malignant transformation, allowing metastases to occur in lymph nodes and other organs.[4,5]

Trichoepitheliomas manifest as normochromic or yellowish papules or firm nodules preferring the central face region, particularly the nose.

Trichoblastomas are rare follicle tumours characterised by solitary nodules with smooth well-defined borders which appear on the head and neck.

In view of the progressive nature of BSS, with recurrence and the risk of malignant transformation, surgical excision is the treatment of choice. Other therapies (dermabrasion, electrodissection, CO_2 laser, cryotherapy, and radiotherapy) are associated with high recurrence rates.[6]

The patient was classified as a BSS carrier by presenting multiple cylindromas, trichoepitheliomas, and trichoblastoma. Surgical excision was programmed, with clinical follow-up and genetic counselling with treatment. ❑

Abstract: Brooke-Spiegler syndrome is an autosomal dominant disorder with variable penetrance and expression. It is characterized by a genetic predisposition to develop multiple adnexal neoplasias: cylindromas, trichoepitheliomas, and trichoblastomas. We describe a 54-year-old male patient with cylindromas, trichoepitheliomas, and trichoblastoma.

Keywords: Neoplasms, adnexal and skin appendage; Skin diseases, genetic; Skin neoplasms

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