# Múltiplas Pápulas na Face

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**PALAVRAS-CHAVE –** Neoplasias Faciais/diagnóstico; Neoplasias da Pele/diagnóstico.



# *Multiple Papules on the Face*

***KEY WORDS –*** *Facial Neoplasms/diagnosis; Skin Neoplasms/diagnosis.*

## CASE REPORT

A 43-year-old female patient was referred to our department due to multiple painless skin colored firm papules and nodules, with a smooth surface, ranging from a few millimeters to 2 cm, located on the face, particularly around the nose and forehead, and scalp, in this latter location causing alopecia. Lesions developed progressively over 12 years (Fig. 1). There were no complaints from other organs and she was under no chronic medication. The patient reported her mother had similar skin lesions that also started around the age of 30.

An excisional biopsy of a skin nodule of the scalp was performed and histological examination showed a dermal lesion composed by multiple lobules arranged in a jigsaw pattern. Each lobule consisted of an outer layer of cells with small hyperchromatic nuclei and an inner zone of cells with oval vesicular nuclei, surrounded by a hyaline mantle (Fig. 2). Genetic study showed a heterozygous nonsense mutation (c.2806C>T, p.Arg936*) of the *CYLD* gene.

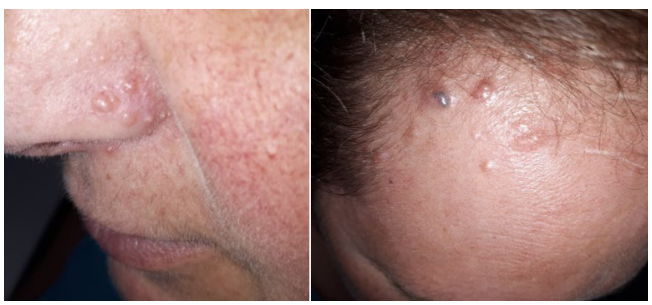

**Figure 1 -** Multiple skin colored papules and nodules located on the face.

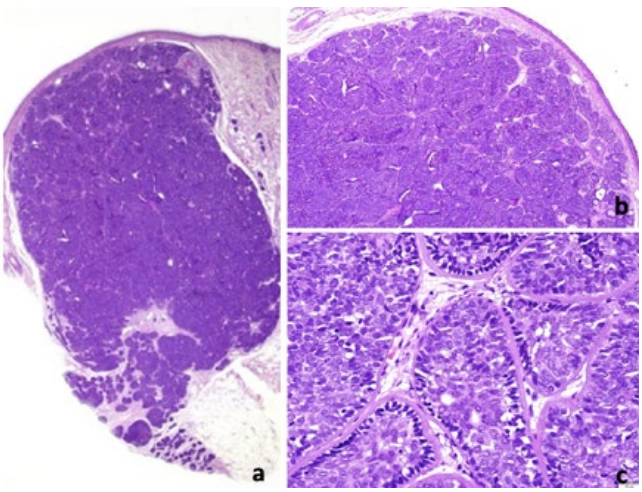


**Figure 2 -** Dermal lesions composed by multiple lobules arranged in a jigsaw pattern typical of cylindroma (image a, H&E, 20x and image b, H&E, 40x). Each lobule consists of an outer layer of cells with small hyperchromatic nuclei and an inner zone of cells with oval vesicular nuclei, surrounded by a hyaline mantle (image c, H&E, 400x).

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# Qual o Seu Diagnóstico?

## WHAT IS YOUR DIAGNOSIS?

### BROOKE–SPIEGLER SYNDROME

Clinical and histological findings combined with family history suggested Brooke–Spiegler syndrome and the genetic test confirmed the diagnosis. Due to the absence of symptomatic complains or aesthetic concerns, the patient refused treatment procedures.

No suspicious lesions were identified during 5 years of follow-up.

Brooke–Spiegler syndrome (BSS) is a rare genodermatosis, with an autosomal dominant pattern of inheritance, caused by mutations in the CYLD gene, a tumor suppressor gene.[1] It is characterized by the development of multiple skin appendage tumors, namely spiradenomas, cylindromas and trichoepitheliomas.[2] Although they are typically benign, malignant transformation occurs in 5% to 10% of the patients.[3] In such cases mostly cylindrocarcinomas develop within the lesions. Less frequently, malignant spiradenomas and basal cell carcinomas were described.[4] Apart from the skin, morphologically similar neoplasms may rarely arise in the salivary glands or breasts (mammary cylindroma).[5]

This case highlights the importance of skin lesions as a diagnostic clue for systemic diseases. A prompt diagnosis enables the genetic counseling of the patient and his relatives, through a multidisciplinary approach, allowing the early detection of the underlying malignancies, namely malignant transformation of cutaneous lesions and salivary glands tumors.[6]

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### REFERENCES

1. Pinho AC, Gouveia MJ, Gameiro AR, Cardoso JC, Gonçalo MM. Brooke-Spiegler Syndrome – an underrecognized cause of multiple familial scalp tumors: report of a new germline mutation. J Dermatol Case Rep. 2015;9: 67-70. doi: 10.3315/jdcr.2015.1208.
2. Manchanda K, Bansal M, Bhayana AA, Pandey S. Brooke-Spiegler syndrome: a rare entity. Int J Trichology 2012; 4: 29–31. doi: 10.4103/0974-7753.96084.
3. Kazakov DV. Brooke-Spiegler Syndrome and phenotypic variants: an update. Head Neck Pathol. 2016; 10:125-30. doi: 10.1007/s12105-016-0705-x.
4. Mohiuddin W, Laun J, Cruse W. Brooke-Spiegler Syndrome. Eplasty. 2018; 18:ic14.
5. Scott AR, Faquin WC, Deschler DG. Parotid mass in a woman with multiple cutaneous cylindromas. Head Neck. 2010;32:684-7. doi: 10.1002/hed.21133.
6. Lavorato FG, Miller MD, Obadia DL, Nery NS, Silva RS. Syndrome in question. Brooke-Spiegler syndrome. An Bras Dermatol. 2014; 89:175-6. doi: 10.1590/abd1806-4841.20142194.