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Basaloid follicular hamartoma

Hamartoma folicular basaloide

Case Report

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ABSTRACT
Basaloid follicular hamartoma (BFH) is a rare and benign adnexal tumor that resembles basal cell carcinoma (BCC) and may present with different clinical manifestations. A mutation in the PTCH gene, involved in Gorlin-Goltz syndrome, could be associated with the pathogenesis of this neoplasm. We describe the case of a 7-year-old girl with multiple papules on her face.
Keywords: Hamartoma; Carcinoma, Basal cell; Genes, Tumor suppressor.

RESUMO
O hamartoma folicular basaloide (HFB) é um tumor anexial raro e benigno, que se assemelha ao carcinoma basocelular (CBC), e pode apresentar manifestações clínicas diversas. Uma mutação no gene PTCH, envolvido na síndrome de Gorlin-Goltz, poderia estar associada à patogênese dessa neoplasia. Descreve-se caso de menina, sete anos, apresentando múltiplas pápulas na face.
Palavras-chave: *Hamartoma; Carcinoma basocelular; Genes supressores de tumor.*

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INTRODUCTION

Basaloid follicular hamartoma (BFH) is a rare benign tumor of hair follicles whose histopathological features can mimic basal cell carcinoma (BCC).[1,2] It can be hereditary or acquired and localized or generalized. Generalized hereditary forms are usually associated with systemic manifestations, which vary according to the subtype.[1,3]

CASE REPORT

A seven-year-old girl, previously healthy, presented papules in the central region of the face for about a year without associated symptoms. There was no relevant family history. The examination revealed mildly hypochromic papules on the nasal dorsum and malar regions **(Figure 1),** with no other skin lesions. The diagnostic hypotheses were milia, sebaceous hyperplasia, and syringoma. We performed an incisional biopsy, whose histopathology showed basaloid cells forming anastomosed cords and palisade distribution, initially suggesting nodular basal cell carcinoma (Figure 2). We opted for an anatomopathological review and immunohistochemistry, which showed positive Bcl-2 in the outermost layer of tumor cells, positive CD-10 in the surrounding stromal cells, Ki-67 index (proliferative activity) of 20%, and CD- 34 negative (Figure 3). Thus, we established the diagnosis of basaloid follicular hamartoma. The patient initiated a thorough investigation to rule out associated syndromes.

DISCUSSION

Although the pathogenesis of BFH is unknown, it is believed that a mutation in the PTCH (protein patched homolog) gene, which encodes a receptor involved in the Sonic hedgehog–Patched–Gli (Shh-Ptch-Gli) signaling pathway, could contribute to the tumor formation. This abnormality would promote an inadequate regulatory function, with constant positive signaling, resulting in atypical and uncontrolled cell division and growth.[3-5]

The clinical manifestations are varied, but most cases of BFH present with one or multiple papules of 1 mm to 2 mm of normochromic to brownish color on the face, scalp, neck, axilla, trunk, and pubic region.[1,5] In this case, the lesions were mildly hypopigmented, multiple, and bilateral.

So far, five clinical forms have been described: (1) Solitary or multiple papules, as in the report; (2) Linear or unilateral localized papule or plaque; (3) Localized plaque with alopecia; (4) Autosomal dominant, generalized familial type, without associated diseases; and (5) Generalized papules associated with myasthenia gravis, alopecia, systemic lupus erythematous, hypotrichosis, and cystic fibrosis.[2,3,5]

Recently, a retrospective study conducted at the University Hospital of Strasbourg, France, evaluating 17 cases of BFH diagnosed between 1998 and 2017, described a higher incidence of the solitary form, characteristically confused with basal cell carcinoma. It also reported that the unilateral linear form often follows Blaschko's lines, reflecting cutaneous mosaicism. This clinical variant may be related to the ipsilateral brain, bone, and dental abnormalities. It may also be associated with Happle--Tinschert syndrome.[4]

Despite the variability of BFH manifestations, the histopathological appearance is remarkably constant. There are cords and extensions of branched basaloid epithelial cells arranged vertically in the superficial and middle dermis, most in connection with the epidermis and hair follicles, as in the case reported. When the palisade organization is present, it is much less marked than the BCC. By definition, cell atypia and mitosis are absent or very rare. Follicular involvement is marked by the vertical arrangement that replaces the normal follicle, lesion with a basophilic periphery and eosinophilic center, and mucin-rich stroma.[1,4]

Immunohistochemistry is not specific but may aid in the diagnosis. In the stroma, there is positivity for CD-34 and CD-

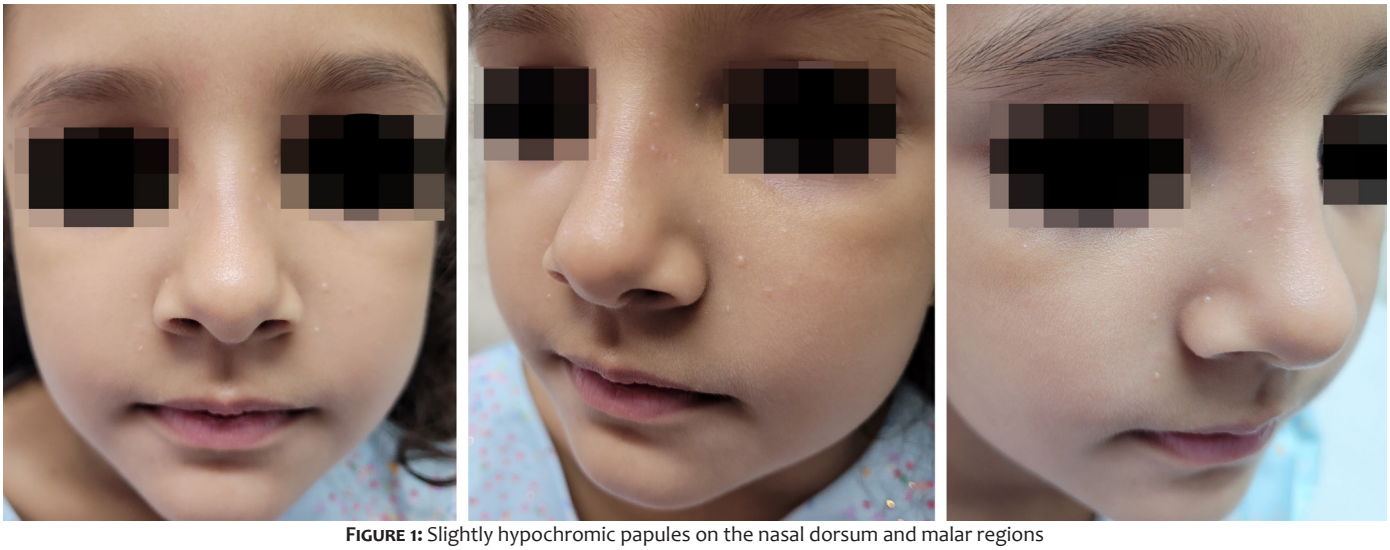

Figure 1: Slightly hypochromic papules on the nasal dorsum and malar regions

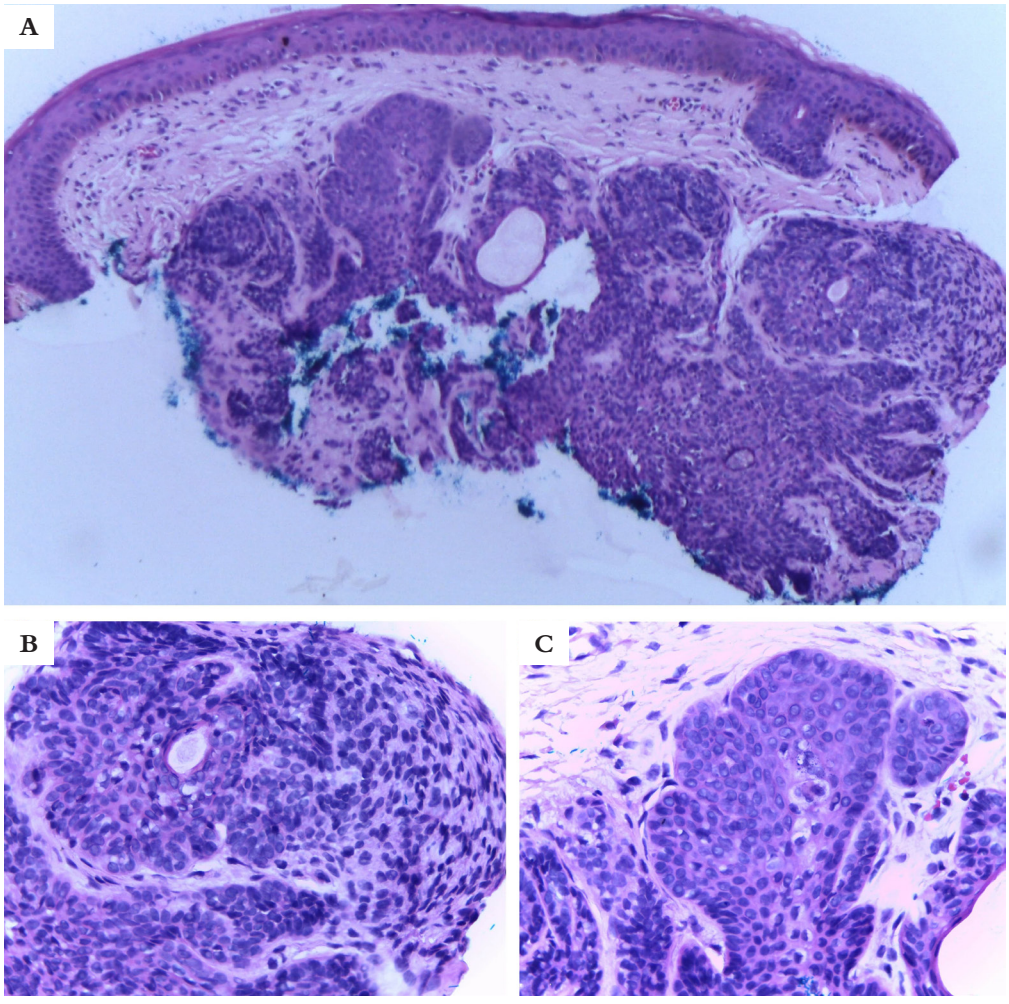

Figure 2: At lower **(A)** and higher **(B and C)** magnifications, basaloid cells forming anastomosed cords and palisade distribution (Hematoxylin & eosin, 40x, 400x, 400x)

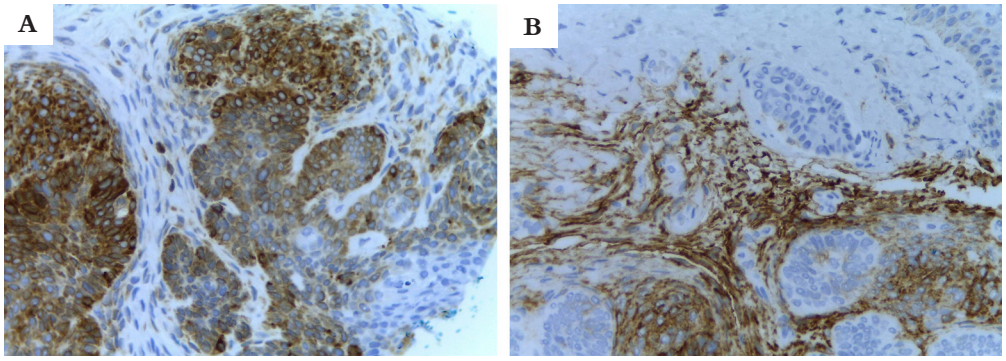

Figure 3: (A) Immunohistochemistry with positive Bcl-2 only in the outermost tumor cells. **(B)** Immunohistochemistry demonstrating CD-10 positivity in stromal cells

10 (which also stains matrix cells). Tumor cells present an immunoreaction for Bcl-2 restricted to a few cells on the periphery of the islands, as in the report. The Ki-67 index is low. PCNA is not very prominent, and PTCH mRNA is overexpressed in cells in direct contact with the dermis, while CD-10 of tumor cells is negative.[2]

The primary differential diagnosis of localized BFH is basal cell carcinoma, especially the infundibulum cyst. Contrary to BFH, BCC immunohistochemistry shows negative CD-34 in the stroma, diffusely positive Bcl-2, prominent PCNA, diffusely overexpressed PTCH mRNA, and positive CD-10, with a high Ki-67 index.[1,2] In cases of multiple localized lesions, as in

the present report, other hypotheses would be adnexal tumors, trichoepitheliomas, sebaceous hyperplasias, syringomas, and sebaceous nevi.

The malignancy potential of BFH is uncertain, although transformation to BCC has been documented. Of the ten transitional cases reported, eight were linear or unilateral localized variants.[2] It is believed that the rapid growth or change in the clinical appearance of the lesions could alert about BCC development.[2-4] Prophylactic excision is not recommended, especially in multiple presentations.[4]

There is still no established algorithm for the BFH treatment. The literature describes options, such as surgical excision, cryotherapy, CO2 laser, photodynamic therapy, and imiquimod. We chose expectant management with regular clinical follow-up, considering the age of the reported patient and the existence of multiple lesions on the face. Vismodegib, an inhibitor of the hedgehog signaling pathway, may help to treat severe conditions in the future. The use of 5-aminolevulinic acid (5-ALA) associated with photodynamic therapy is safe, being a possible therapy for children with multiple lesions.[1-5]

The prognosis of basaloid follicular hamartoma is excellent, with a notable exception for cases associated with the development of BCC or systemic disorders.[3]

CONCLUSION

Basaloid follicular hamartoma is a rare benign neoplasm easily confused, clinically and histopathologically, with BCC. We present the case of a seven-year-old girl with multiple papules in the malar region and on the nasal dorsum, whose initial anatomopathological examination suggested BCC. It is crucial to consider the BFH in the presence of multiple nonspecific lesions in the pediatric age group in healthy children. In this case, revisions of the histopathological study and immunohistochemistry were essential for the diagnosis. ●

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