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Basaloid Follicular Hamartoma Associated with Brooke-Spiegler Syndrome

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Abstract **Case Report**

Basaloid follicular hamartoma is a benign, superficial malformation of hair follicles that can be mistaken both clinically and histopathologically for basal cell carcinoma. Basaloid follicular hamartoma has been described as a solitary lesion or with other genodermatoses such as Bazex-Dupre-Christol syndrome, BCNS. Here we present a 73-year-old patient with an asymptomatic congenital lesion on the forehead, scalp, and facial which increased in size over the years. Histopathology showed a basaloid follicular hamartoma associated with the complete phenotype of Brooke-Spiegler syndrome, an association that is rarely described.

Keywords: Basaloid follicular hamartoma, Brooke-Spiegler syndrome, Cylindroma, Trichoepithelioma, Spiradenoma, CYLD gene.

INTRODUCTION

Basaloid follicular hamartoma (BFH) is a rare benign adnexal neoplasm characterized by a histological appearance that closely mimics both infundibulocystic basal cell carcinoma and trichoepithelioma [1]. It may occur as a familial or sporadic condition and can present in generalized or localized forms [2]. In some cases, BFH is associated with mutations in the PTCH gene [3]. Given its variable clinical presentation, a skin biopsy is essential to distinguish BFH from other common cutaneous lesions [1].Brooke-Spiegler syndrome (BSS) is a rare autosomal dominant genodermatosis caused by mutations in the CYLD gene [4], characterized by a predisposition to develop multiple benign adnexal tumors, classically represented by the triad of cylindromas, trichoepitheliomas, and spiradenomas, with a highly variable phenotype lesions [5]. We report here a case of Brooke-Spiegler syndrome in a 73-year-old patient, with a phenotype including a basaloid follicular hamartoma, rarely described in the literature.

CASE REPORT

A 73-year-old man presented with multiple cutaneous lesions on the scalp and face. The lesions first appeared at age 20, initially on the face, then spread to the scalp, gradually increasing in size and number over the years. His father had similar facial lesions. On clinical examination, multiple round-to-oval, skin-colored papules and nodules with a smooth, pearly surface were observed on the mid-face, particularly in the nasolabial folds, periocular areas, and less commonly on the lower lip and chin (Figure 1a). On the scalp, there were two distinct lesions. The larger one, located on the vertex, was a firm, pinkish-red, dome-shaped tumor measuring 1.5 cm, completely hairless, with obvious surface telangiectasia and induration. The smaller one, near the frontal hairline, was a pink to bluish nodule (Figure 1). Dermoscopy of the facial papules demonstrated arborizing vessels, shiny white areas, and a yellowish-brown background, with structureless white-yellow areas noted in the chin lesion. Dermoscopy of the vertex tumor displayed prominent arborizing vessels and a pink-orange homogeneous background. The frontal lesion showed a light blue-gray background with branched vessels (Figure 2). The patient was otherwise in good general health, with no known underlying medical conditions or prior medication use. Histopathological examination of the larger scalp nodule revealed a well-circumscribed dermal tumor with cribriform architecture composed of basaloid cells with scant cytoplasm, homogeneous chromatin, and separated

by thickened hyaline basement membrane material, consistent with cylindroma. Facial papules showed a dermal proliferation connected to the epidermis, organized into basaloid masses centered by keratinized cysts, with anastomosing trabeculae in some areas. Peripheral palisading was present. Although rare mitoses were observed, the absence of retraction clefts and the highly focal myxoid stroma made distinction from basal cell carcinoma difficult. Immunohistochemistry with negative Bcl-2 and CD10 within tumor cells, very limited p53 expression, ruled out basal cell carcinoma and confirmed trichoepithelioma (Figure 3). A lesion in the mental region demonstrated numerous cords and strands of basaloid cells arranged in a radial and anastomosing pattern over scant fibrous stroma, consistent with basaloid follicular hamartoma. A forehead nodule showed sheets of basaloid cells with paler central nuclei and sparse cytoplasm, consistent with spiradenoma. Based on the clinical presentation, dermoscopic findings, and histopathological features, a diagnosis of Brooke-Spiegler syndrome (BSS) with associated basaloid follicular hamartoma was established.

Figure 1: Multiple round-to-oval, skin-colored papules and nodules on the mid-face, particularly in the nasolabial folds, the lower lip, and chin(A). Pinkish-red, dome-shaped tumor, with surface telangiectasia(B). Pink to blueish nodule near the frontal hairline(C)

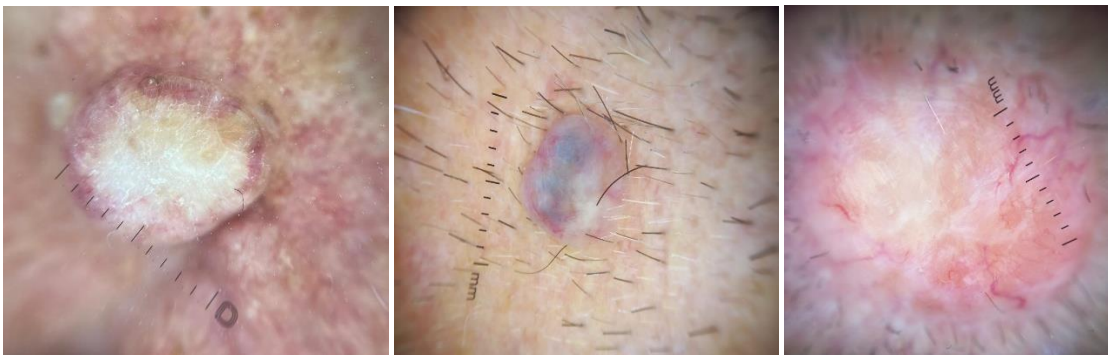

Figure 2: Dermoscopy of the facial papules shows arborizing vessels, shiny white areas, with structureless white-yellow areas(A). Dermoscopy of the vertex tumor reveals prominent arborizing vessels and a pink-orange homogeneous background(B). Dermoscopy shows a light blue-gray background with branched vessels(C)

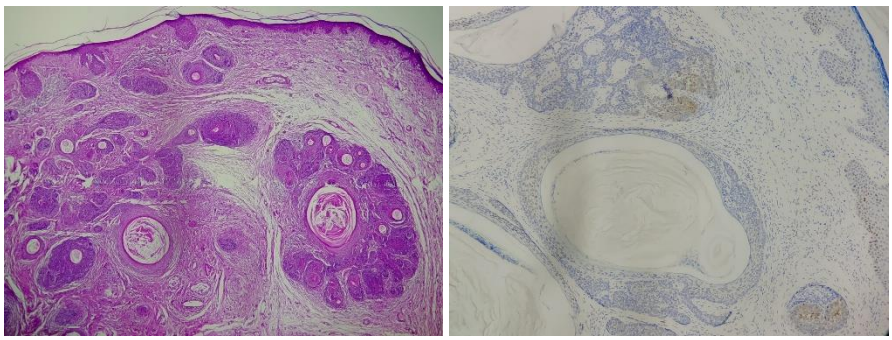

Figure 3: Trichoepithelioma (cribriform/trichoblastoma variant). Punch biopsy from the face demonstrating a well-circumscribed dermal proliferation connected to the overlying epidermis. The tumor consists of multiple basaloid cell aggregates arranged in lobules and anastomosing trabeculae, with numerous keratin-filled horn cysts within the tumor nests. Peripheral palisading is evident. The stroma is fibrous with focal myxoid areas and no retraction clefts. Hematoxylin and eosin (H&E), ×100 (A). Immunohistochemistry with negative Bcl-2 and CD10 within tumor cells, very limited p53 expression(B)

DISCUSSION

Basaloid follicular hamartoma (BFH) is a rare, benign adnexal neoplasm originating from hair follicle epithelium [6]. It was first described by Brown et al. in 1969 and formally named by Mehregan and Baker in 1985 [3]. Basaloid follicular hamartoma may be congenital or acquired [2]. Its clinical presentation is highly variable, typically manifesting as skin-colored to brown papules, nodules, or plaques, most commonly on the face and scalp, and occasionally on the trunk [3]. Five distinct clinical forms have been described: Solitary or multiple papules, localized linear or unilateral plaque, often distributed along Blaschko's lines, which may be present at birth or develop during childhood, localized plaque with alopecia [1], generalized familial type, without associated systemic disorders, generalized acquired or congenital form associated with diffuse alopecia and various conditions, including myasthenia gravis, systemic lupus erythematosus, cystic fibrosis, or other ectodermal defects [6]. BFH may occur as an isolated finding or in association with other hereditary syndromes, such as Bazex-Dupré-Christol syndrome, Gorlin syndrome, or Happle-Tinschert syndrome [7]. Despite their variable clinical presentations, all forms of basaloid follicular hamartoma (BFH) share highly consistent histopathological features: multifocal islands and branching cords of basaloid epithelial cells in the papillary dermis, often connected to the epidermis or dilated hair follicles, with frequent keratin cysts within the cords or lace-like networks. Cellular atypia and mitoses are absent [8]. Immunohistochemically, BFH shows CD34 positivity in the stroma and peripheral Bcl-2 expression in the basaloid trabeculae, with a low Ki67 index. These features distinguish BFH from its main mimics, particularly infundibulocystic basal cell carcinoma (BCC) [1]. PTCH, a tumor suppressor gene encoding a transmembrane receptor, is frequently mutated in basaloid epithelial neoplasms such as trichoepithelioma [6]. Although basaloid follicular hamartoma (BFH) may also carry PTCH mutations, these are both rarer and distinct from those in more malignant basaloid lesions, which aligns with the consistently benign behavior of BFH [9].

The Brooke-Spiegler syndrome (BSS) is a rare autosomal dominant genodermatosis with variable penetrance that predisposes affected individuals to multiple adnexal neoplasms [10]. Clinically, it is characterized by the simultaneous or sequential development of numerous benign adnexal tumors, most classically cylindromas, trichoepitheliomas, and occasionally spiradenomas [11]. BSS results from mutations or loss of heterozygosity in the cylindromatosis gene (CYLD) located in chromosome 16q12-q1[12]. The first tumors usually appear at puberty, with a slight female predominance, and their number and size gradually increase throughout life [10], as was the case with our patient. Trichoepithelioma represents a neoplasm with follicular germinative differentiation; it manifests as flesh colored or yellowish papules or firm nodules, preferring the central face region, particularly the nose [13]. Dermoscopically, it is characterized by small, thin arborizing vessels, shiny white areas/background, and milia-like cysts (keratin cysts on histology),yellowish-brown background color [7] as seen in our case. Cylindroma is a benign adnexal tumor that predominantly affects the scalp and head, typically presenting as slowly growing, solitary pink to red dermal nodules [14]. Spiradenoma is also a benign neoplasm, usually appearing as a solitary, intradermal, round-to-oval lesion that is pink or blue in color [15]. In dermoscopy, both tumors share similar features, most commonly showing a homogeneous pink or pink-orange background with prominent arborizing telangiectasia. In some cases, blue ovoid nests or blue globules may also be observed [7]. The frequent coexistence, within the same lesion, of histological features belonging to two or even all three tumor types, cylindroma, spiradenoma, and trichoepithelioma, along with their shared embryologic origin from the folliculosebaceous apocrine unit, supports an apocrine derivation for both cylindromas and spiradenomas [16]. Consequently, Brooke-Spiegler syndrome can be understood as a genetic disorder affecting the regulation and differentiation of the folliculosebaceous-apocrine unit [10]. Malignant transformation remains the most serious concern, especially cylindrocarcinoma in multiple scalp cylindromas [17], and BFH should be closely monitored as they may evolve into BCC [1]. Management is challenging due to the multiplicity and recurrence of tumors; wide local excision remains the gold standard, with dermabrasion, laser (CO2 or erbium:YAG), electrodessication, or cryotherapy as alternatives, depending on location and tumor burden [1-17].

CONCLUSION

In summary, BSS exhibits significant clinical and histological heterogeneity. The coexistence of a complete phenotypic spectrum and basaloid follicular hamartoma, as observed in this patient, is uncommon. Further studies are warranted to better elucidate the phenotypic variability and its underlying genetic mechanisms.

REFERENCES

1. Edelman, S., Torres Huamani, A. N., Centeno, M. D. V., & Cervini, A. B. (2022). Basaloid follicular hamartoma associated with follicular mucinosis and inflammation. *Anais Brasileiros de Dermatologia, 97*(1), 45–48.
2. Huang, S., Hsiao, T., & Lee, C. (2011). Basaloid follicular hamartoma: A case report and review of the literature. *The Kaohsiung Journal of Medical Sciences, 28*(1), 57–60.
3. Tiodorovic, D., Jovic, A., Cekic, S., Vidovic, N., Radevic, T., & Mijušković, Ž. (2024). Solitary

basaloid follicular hamartoma: A report of two cases and a brief review of the literature. *Vojnosanitetski Pregled, 81*(1), 11–11.

4. Tse, J. Y., & Hoang, M. P. (2012). A review and update of skin appendage neoplasms and associated genetic syndromes. *Expert Review of Dermatology, 7*(3), 235–245.
5. Kazakov, D. V. (2016). Brooke-Spiegler syndrome and phenotypic variants: An update. *Head and Neck Pathology, 10*(2), 125–130.
6. Mauleón, C., Valdivielso, M., Chavarría, E., Hernanz, J. M., & Casado, I. (2011). Simultaneous presentation of localized basaloid follicular hamartoma and epithelioid blue nevus in a 44-year-old patient. *Actas Dermo-Sifiliográficas, 102*(3), 233–235.
7. Zaballos, P., Gómez-Martín, I., Martin, J. M., & Bañuls, J. (2018). Dermoscopy of adnexal tumors. *Dermatologic Clinics, 36*(4), 397–412.
8. Jain, S., Khopkar, U., & Sakhiya, J. (2019). Localized unilateral basaloid follicular hamartoma along Blaschko's lines on face. *Indian Journal of Dermatology, Venereology and Leprology, 85*(1), 60.
9. Jih, D. M., Shapiro, M., James, W. D., Levin, M., Gelfand, J., Williams, P. T., Oakey, R. J., Fakharzadeh, S., & Seykora, J. T. (2003). Familial basaloid follicular hamartoma: Lesional characterization and review of the literature. *The American Journal of Dermatopathology, 25*(2), 130–137.
10. Mataix, J., Bañuls, J., Botella, R., Laredo, C., & Lucas, A. (2006). Síndrome de Brooke-Spiegler: una entidad heterogénea. *Actas Dermo-Sifiliográficas, 97*(10), 669–672.
11. Portincasa, A., Cecchino, L., Trecca, E. M. C., Lembo, F., Annacontini, L., Ciancio, F., Corsi, F., Cassano, M., & Parisi, D. (2018). A rare case of Brooke-Spiegler syndrome: Integrated surgical treatment of multiple giant eccrine spiradenomas of the head and neck in a young girl. *International Journal of Surgery Case Reports, 51*, 277–281.
12. Lavorato, F. G., Miller, M. D. B., Obadia, D. L., Nery, N. S., & Silva, R. S. da. (2014). Syndrome in question. *Anais Brasileiros de Dermatologia, 89*(1), 175–176.
13. Mohammadi, A. A., & Seyed Jafari, S. M. (2014). Trichoepithelioma: A rare but crucial dermatologic issue. *World Journal of Plastic Surgery, 3*(2), 142–145.
14. Alwakaa, O., Abo Saada, S., Eid, H., & Etr, A. (2025). Cylindroma of the neck in a male patient: A case report. *Journal of Medical Case Reports, 19*(1), 554.
15. Dhua, S., & Sekhar, D. (2015). A rare case of eccrine spiradenoma—treatment and management. *European Journal of Plastic Surgery, 39*(1), 1–4.
16. Fenske, C., Carter, N., Banerjee, P., & Holden, C. (2000). Brooke–Spiegler syndrome locus assigned to 16q12-q13. *Journal of Investigative Dermatology, 114*(5), 1057–1058.
17. Layegh, P., Sharifi-Sistani, N., Abadian, M., & Moghiman, T. (2008). Brooke-Spiegler syndrome. *Indian Journal of Dermatology, Venereology and Leprology, 74*(6), 632.