CASE REPORT

Basaloid follicular hamartoma syndrome: acquired sporadic variant with hypothyroidism, hypohidrosis and alopecia, a rare case

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Abstract

Basaloid follicular hamartoma is a rare benign malformation of hair follicles, characterised clinically as generalised or localised multiple brown papules mostly on face, scalp and trunk. It may be congenital or acquired with or without any associated disease. Histologically it is composed of epithelial proliferation of basaloid cells with radial disposition enclosed in a fibrous stroma. It is of important consideration because it can be mistaken for basal cell carcinoma both clinically and histologically. Here we report the case of a 51-year-old female with acquired, generalised basaloid follicular hamartomas associated with alopecia, hypothyroidism and hypohidrosis which is an extremely rare disease.

Keywords: Basaloid follicular hamartoma, benign neoplasm, basal cell carcinoma, rare disease, nevoid basal cell carcinoma syndrome.

DOI: 10.47391/JPMA.6560

Submission completion date: 18-04-2022
Acceptance date: 22-12-2022

Introduction

Basaloid follicular hamartoma (BFH) is a rare benign superficial malformation of the hair follicles associated with epithelial proliferation of the basaloid cells, perhaps an abortive hair growth of secondary hair germs with a limited differentiation towards the upper follicular portion.[1,2] Currently it has 5 described clinical forms: solitary papule, nodule, or plaque; localised linear or unilateral plaque; generalised autosomal dominant without associated disorders; generalised papules associated with hereditary skin disease such as nevoid basal cell carcinoma syndrome; generalised papules (acquired form) with associated disorders such as lupus erythematous, myasthenia gravis, alopecia, cystic fibrosis etc.[3,4] Mutation in patch (PTCH) gene located on chromosome 9q23 causes BFH and nevoid basal cell carcinoma syndrome. Expression of this mutation is milder in BFH, as these are benign tumours. Due to its rare chances of malignant transformation, skin biopsy should not be delayed.[5] Histopathologically it is composed of multiple cords and strands of anastomosing basaloid cells with follicular differentiation originating from the infundibular portions of the hair follicles.[6] Management includes observation for any malignant changes and removal of lesions mainly for cosmetic disfigurement by surgical excision, cryosurgery, photodynamic therapy or lasers such as ruby, alexandrite, Nd-Yag and CO_2. The prognosis is usually good, unless BCC or associated systemic disorders develop.[7] We report the case of a female with acquired generalised BFH which developed sporadically. To our knowledge, this is the first case being reported from Pakistan. This aims to highlight the rarity yet significance of this disease in resemblance to infundibulocystic basal cell carcinoma (BCC).

Case Report

A 51-year-old female patient who presented to the Dermatology Out Patient Department of PNS Shifa Hospital on 11-02-2021 with history of numerous generalised brownish papules, hair fall and decreased sweating for 1.5 years. She was a known case of hepatitis B for the past 11 years and was taking Tenofovir 300mg once daily since 2014. There was no history of myasthenia gravis (MG), or any other associated autoimmune disease nor history of such lesions in the family. Physical examination revealed multiple tan-skin coloured homogenous papules 1-5mm on head, neck, limbs and trunk with highest concentration on the scalp, back of the neck, upper chest and upper back with no coalescence as shown in figure 1. There was hyperpigmentation of the tongue and buccal mucosa with diffuse thinning of scalp hair as shown in figure1. Her nails, palms and soles were spared. Rest of the systemic examination was also unremarkable. Laboratory investigations, including complete blood count, liver and renal function test and fasting blood glucose were within normal range. Her thyroid profile showed raised TSH levels (10.15 IU/ml, normal range 0.5-8.9 IU/ml), decreased free T4 levels (0.83ng/dl, normal range 0.9-2.3ng/dl) with negative Anti-TPO antibodies. For this she was prescribed tab levothyroxine 50mcg once daily by Endocrinologist.

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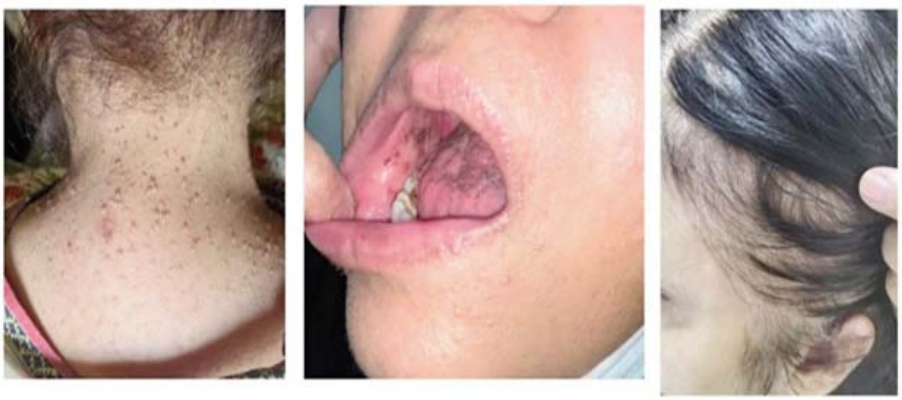

Figure-1: Multiple skin coloured papules on back of neck, B. hyperpigmentation of the tongue and buccal mucosa, C. Alopecia.

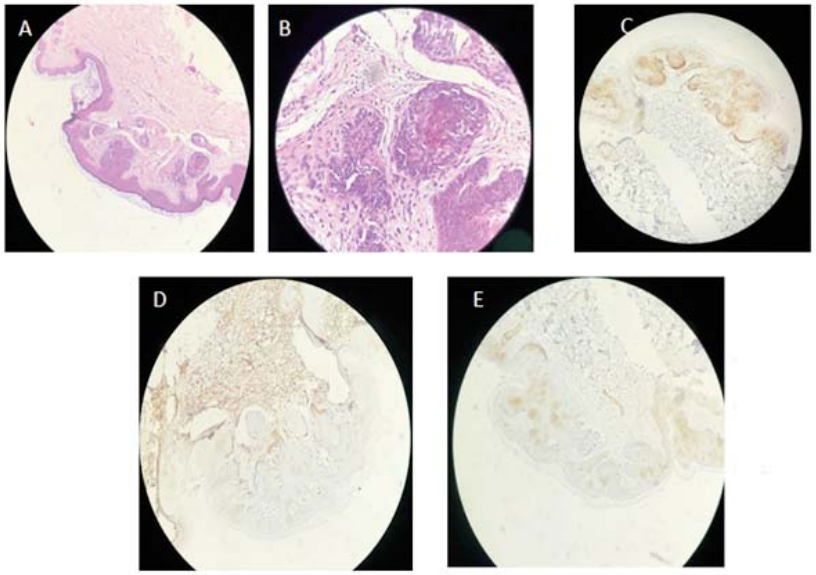

Figure-2: A. Histopathology of BFH showing basaloid and squamous cells in superficial dermis (x10). B. Anastomosing strands of basaloid cell (x40) C. Immunostaining showing positivity for Bcl-2, D. CD34 & E. EMA (x40)..

Serum ANA profile, anti ds-DNA, ultrasound abdomen, skeletal survey were normal. In our patient, the differentials were multiple trichoepitheliomas, trichilemmoma, eruptive syringoma and papular drug eruption with eosinophilia (rare side effect of tonofovir). A skin biopsy from the back of neck was taken under local anaesthesia and was sent for histopathological examination. It showed anastomosing strands of basaloid and squamous cells in superficial dermis with no attachment to the overlying epidermis. There were thick collagen bundles and mild perivascular lymphocytic infiltrate with no atypical cellular features or mitotic activity. Immunohistochemistry was positive for CD34, Bcl-2 and EMA. Bcl-2 was positive only in outermost cells adjacent to the stroma as shown in figure 2.

Thus, a diagnosis of acquired multiple BFH syndrome with hypothyroidism, hypohidrosis and alopecia was made. The patient was counselled regarding the benign nature of her skin lesions with treatment options of CO2 laser ablation and excision of lesions but she preferred to remain under observation with 3 monthly follow up visits as advised. Number of lesions have gradually increased but none of the lesions look suspicious.

Discussion

BFH presents in a variety of clinical forms. Generalised BFH syndrome presents with pappules accompanied by alopecia, myasthenia gravis, systemic lupus erythematosus, hypohidrosis or cystic fibrosis rarely.[4] El-Darouti et al reported generalised variant of BFH with no associated internal disorders.[8] The patient in our case presented with acquired multiple generalised BFH with alopecia, hpothyroidism and hypohidrosis. Our patient also had hyperpigmentation of the tongue and buccal mucosa which has not been previously reported. The histopathology is highly useful to confirm the diagnosis. The stromal cells of the BFH stain was positive for CD34, but the stromal cells of BCC were negative. Bcl-2 stains only the outermost tumour cells of BFH but is more prominent in BCC.[9] In our case, histopathology showed similar changes. Cerejeira et al treated BFH with CO2 laser with satisfactory results.[10] In our case the patient was also advised ablation of lesions with CO2 laser but she opted non-interventional treatment. Our patient has been taking Tenofovir for 08 years. Tenofovir associated cutaneous adverse effects are rare and compasses maculopapular or vesiculopustular rash, urticaria, hypersensitivity reaction, lichenoid drug eruption, photoallergic dermatitis and bullous pemphigoid lesions. The opinion of a Gastroenterologist was sought, who advised to continue the treatment. Therefore we could not ascertain its association with BFH. Thus, our case also highlights that tenofovir can also be a risk factor for BFH but this needs further research.

Although BFH classically remains stable for years, Gumaste et al reported development of BCC in BFH.[3] Therefore our patient was followed up on 3 monthly basis.

Conclusion

BFH is a benign neoplasm, however, its importance lies in the similarity with BCC both clinically and histologically therefore a biopsy should never be delayed. Although it runs a benign course but the risk of malignant transformation to BCC needs vigilant follow-up.

Patients' Consent: : Informed consent was obtained from patient for the writing of the manuscript and the use of data for research purposes.

Disclaimer: None.

Conflict of Interest: None.

Source of Funding: None.

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