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Multiple Familial Trichoepithelioma: A Rare Case Report Spanning Three generations

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Abstract

Multiple familial trichoepithelioma (MFT) is an uncommon, benign, adnexal tumor with an autosomal dominant inheritance pattern. It manifests as multiple, tiny, firm, skin-colored papules typically on the face. This case report describes five affected individuals across three generations, highlighting the clinical features, diagnostic challenges, and management considerations. Despite its benign nature, the condition carries significant cosmetic and psychological implications. Histopathology remains the gold standard for diagnosis, distinguishing it from malignant mimickers. Various treatment options have been explored, including surgical excision, laser therapy, radiofrequency ablation, topical agents like imiquimod, and electrosurgical techniques, with varying degrees of success. However, recurrence remains a challenge, and treatment depends on the degree of the disease and patient preference. Given the rarity of MFT, increasing awareness among clinicians is crucial for early recognition and appropriate management. This report emphasizes the importance of differentiating trichoepithelioma from other cutaneous neoplasms to prevent unnecessary interventions and optimize patient outcomes.

Key words: Adnexal tumour, Multiple familial trichoepithelioma, Skin appendage neoplasm.

Introduction

Trichoepithelioma is a relatively rare, benign tumour of the hair follicle unit that arises from hair germ portion of hair follicles.[1] It is further classified into three subcategories- Desmoplastic trichoepithelioma, Multiple Familial Trichoepithelioma and Isolated non-Hereditary trichoepithelioma.[2] MFT also referred to as Brooke and Fordyce disease was first described by Brooke and Fordyce in 1892 as 'Multiple benign cystic epithelioma' and 'Epithelioma adenoids cysticum'.[3] It is an autosomal dominant disease that may occur independently or as a part of Brooke-Spiegler syndrome, which features inherited adnexal neoplasms such as trichoepitheliomas, spiradenomas and cylindromas.[4] There are five types of trichoblastomas i.e. nodular, retiform, columnar and cribriform, of which cribriform trichoblastoma (trichoepithelioma) is the most common.[5] Here we present a case report of multiple familial trichoepitheliomas.

Case report

A 26-year-old female presented with multiple, painless, non-pruritic skin coloured papules found primarily over the central aspect of face, nose, forehead and few over the scalp. She first developed lesions at the age of 11 years

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which progressed to the whole face over the next 2 years. Lesions have continued to emerge till date. There were no other cutaneous lesions found on the body. Systemic examination was normal. Similar lesions were reported in her grandmother, mother and two siblings. No children in the subsequent generations similar lesions at present.

Figure 1(a) and (b): Multiple centro-facial papules and nodules seen in the patient

Dermatological examination revealed, multiple, well-defined, clustered and few discrete, skin colored to yellowish, dome shaped papulo-nodular lesions which had a smooth, shiny surface, with firm consistency and of size ranging from 1.5-5mm located over the nose, nasolabial folds and forehead [Figure 1(a)]. Few discrete lesions were present over the scalp, chin and both cheeks. [Figure 1(b)]. Dermoscopy showed multiple arborizing pattern of vessels, multiple milia like cysts and rosettes over a white backdrop [Figure 2].

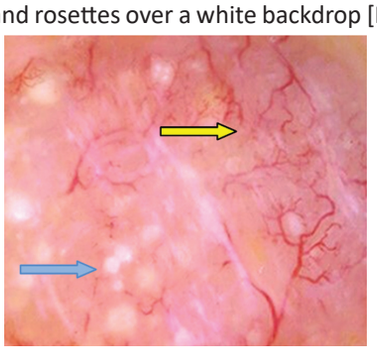

Figure 2: Dermoscopy with polarized light 10X showing multiple milia like cysts(blue arrow) and multiple arborizing vessels (yellow arrow)

A family history of similar lesions in sister and mother was also reported. [Figure 3(a,b)] [Figure 4]

Figure 3(a,b): Multiple trichoepitheliomas over the face in patient's sister and mother

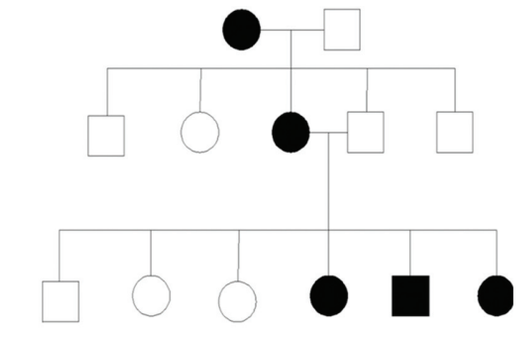

Figure 4: Family tree; five members of the family affected by trichoepithelioma.

Histopathology displayed multiple islands of dark, basaloid epithelial cells with peripheral palisading and surrounding fibrous stroma. No artefactual retractions were seen. [Figure 5]

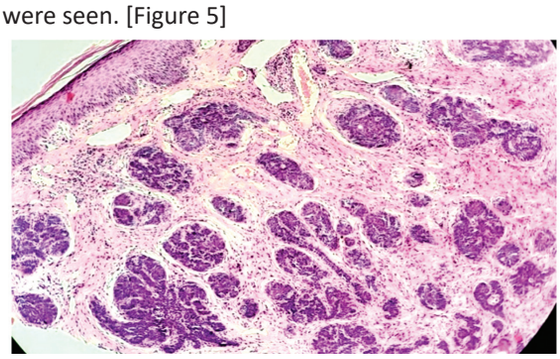

Figure 5 : Histopathology (Hematoxylin and Eosin stain 40X): Areas of small, dark basaloid cells with peripheral palisading, surrounding a central area of eosinoplilic amorphous material.

Discussion

MFT, due to its autosomal dominant pattern of inheritance affects both men and women. However, females are more likely to be affected due to increased gene expression.6 MFT is attributed to two gene mutations, the first one located on chromosome 9p21 and second, a mutation in the CYLD tumour suppressor gene discovered on chromosome 16q12-13.[4,7,8] CYLD protein inhibits the activation of nuclear factor kB(NFkB). NFkB plays a role in inflammation, immunological response, oncogenesis and preventing apoptosis. Inhibiting CYLD causes prolonged cell survival thus leading to tumorigenesis.[9,10] MFT is hypothesized to be a phenotypic variation of Brooke-Spiegler syndrome as it is also associated with the CYLD gene.[11] Trichoepitheliomas can be found in other rare syndromes such as the Rombo syndrome and Basex syndrome.[5]

Hereditary forms of trichoepithelioma usually presents in younger age between 10 to 20 years as multiple, skin coloured, firm, rounded, translucent, shiny, papules or nodules of size ranging from 2-6mm with a umbilicated center.[12] They are typically clustered symmetrically in the centrofacial aspect primarily over the nasolabial

fold, nose, eyelids and face, forehead and eyelids and may occasionally extend to the neck, scalp and upper trunk. The papules gradually increase in size and number over time causing cosmetic disfigurement and psychological impact.[10] Lesions are commonly asymptomatic, however the appearance of symptoms such as itching or ulceration should raise suspicion of Basal cell carcinoma.[6]

The tumour nests contain a large number of Merkel cells and CD34 positivite dendrocytes, indicating an etiology from hair structures. Trichoepitheliomas share histological similarities with Basal cell carcinoma (BCC), especially the solitary variant.[12] The following features support the diagnosis of trichoepithelioma over BCC-smooth, symmetrical lesions, clefts between stroma, lack of artefactual retraction between the tumour cells and the surrounding stroma, fibrocytic instead of mucinous stroma, well developed cornified cysts and papillary mesenchymal bodies, mature follicular differentiation and low mitotic activity.2,6 Dermoscopy in trichoepithelioma shows a pale white background with milia-like cysts and arborizing vessels which matched the findings of our case.[13,14]

Trichoepithelioma should be clinically differentiated from tricholemmoma, syringoma, colloid milium, trichofolliculoma and histopathologically from microcystic adnexal carcinoma and basaloid follicular hamartoma.[1,8]

Despite being asymptomatic, affected individuals mostly desire treatment for aesthetic concerns since the lesions are extensive and disfiguring which could cause social ostracization. Inappropriate approach to treatment can lead to inadequate therapy and recurrence, while over- treating may cause functional and cosmetic impairment.[15] Topical treatment options such as 5% imiquimod cream 5-7 times a week, sirolimus 1% cream and tretinoin 1% cream have been recommended as an effective treatment options.[16,17] Studies showed the successful treatment of trichoepithelioma using topical 2.5% benzoyl peroxide.[18,19,20] Ablative modalities include surgical excision, punch biopsy, chemical cauterization, cryotherapy, electrosurgery and dermabrasion.[21,22] Laser resurfacing using Erbium:Yag, Argon and CO2 lasers may be employed.[23,24] Systemic therapy such as Aspirin 325mg BD daily along with Adalimumab subcutaneous injection 40mg every other week for the first 2 months, followed by 40 mg every week for the next 8 months to prevent the inhibition of CYLD and the NF-kB induced tumorigenesis has shown promising results.[22] In our patient excision with radiofrequency was done with an acceptable cosmetic outcome.

Genetic analysis was not performed, which could have provided additional insights into the hereditary nature of the disease. Long-term follow-up data on disease progression, recurrence, and psychosocial impact could not be extensively documented. Additionally, while various treatment options were discussed, their long-term efficacy in these patients was not evaluated.

Conclusion

This case report discusses multiple familial trichoepithelioma across three generations, emphasizing its hereditary nature and the need for early diagnosis to distinguish it from similar adnexal tumors. Although benign, it can have significant cosmetic and psychological impact. The report underscores the role of dermatologists in recognizing familial patterns and providing long-term management and counseling.

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