Journal of Pakistan Association of Dermatologists. 2021;**31(3)**:546-549.

Case Report

Multiple familial trichoepitheliomas: A rare autosomal dominant skin tumor

Saba Latif, Arfan ul Bari

Department of Dermatology, PNS Shifa Hospital, Karachi.

Abstract Multiple familial trichoepithelioma (MFT) is a rare autosomal dominant skin disease that presents as multiple small tumors, predominantly on face. The lesions resemble other types of skin diseases that present with papules and nodules. We present a patient with persistent eruptions of asymptomatic but cosmetically disfiguring papulonodular lesions on her face which histologically proved as lesions of trichoepitheioma. There was history of similar lesions in other family members too. She was managed with serial electrocautery, chemical cautery and medium depth chemical peel sessions.

Key words
Familial, trichoepithelioma, basal cell carcinoma, benign facial tumors, appendageal tumors, autosomal dominant.

Introduction

Trichoepithelioma is a rare autosomal dominant condition. The lesions are skin colored, firm papules or nodules of 2-8mm in diameter, slow growing and commonly seen in childhood and adulthood.[1] Clinically, it presents as multiple tumors with pilar differentiation, occurring predominantly on face. The first locus has been previously mapped to chromosome 9P21, but no gene for MFT has been identified to date.[2] Trichoepithelioma may be divided into multiple familial trichoepithelioma, solitary trichoepithelioma and desmoplastic trichoepithelioma. Solitary tumors appear as pale skin colored papules or nodules that can reach 2 centimeters in diameter. Due to its autosomal dominant fashion, both genders receive the gene equally, but because of lessened expressivity and penetrance in males, it is more commonly seen in females.[3] Various mutations in CYLD gene, a tumor suppression gene located on chromosome 16Q12-Q13, have been associated with inherited disorders characterized by multiple adnexal tumors such as trichoepitheliomas, cylindromas, and spiradenomas namely, Brooke-Spiegler Syndrome (BSS), Multiple Familial Tricho-epithelioma, and Familial Cylindromatosis. A number of reports have been published claiming that immunohistochemical stains such as BCL-2 and CD 34 that will help to distinguish basal cell carcinoma from trichoepithelioma. MFT and familial Cylindromatosis are considered phenotypic variants of BSS.[1-4]

Histologically, trichoepitheliomas consists of nests of basaloid cells with follicular differentiation embedded within tumor stroma that dissects through the collagen into the upper reticular dermis. Basal cell carcinomas can have a similar microscopic appearance. Contrasting histologic features are said to include papillary mesenchymal bodies which is a feature of trichoepithelioma but the presence of clefting between the epithelial and stromal components

Address for correspondence
Dr. Saba Latif
Department of Dermatology,
PNS Shifa Hospital, Karachi.
Email: sabalatif98@gmail.com

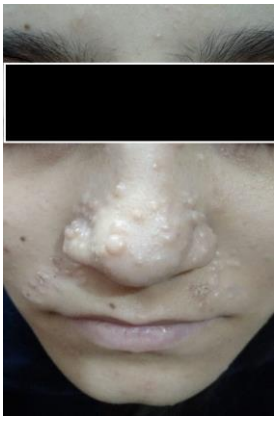

Figure 1a Photograph of patient with multiple facial papules and nodules.

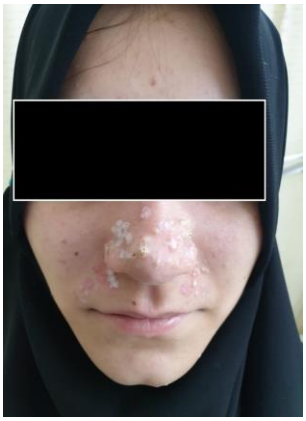

Figure 1b: Photograph of the same patient after electrocautry and 50% TCA.

of the tumor, peripheral palisading of keratinocytes, single cell necrosis, and brisk mitotic rate are associated with basal cell carcinoma and not ordinarily seen in trichoepitheliomas.[4-5] The spindle shaped stromal cells surrounding the tumor nests show strong focal positivity for CD34 while in case of basal cell carcinoma these cells are negative for CD34.[6] Other differentials are molluscum contagiosum, milia, miliaria, syringomas and steatocytoma. Treatment is mainly for cosmetic disfigurement. Various treatment modalities include surgical excision, chemical cauterization, laser resurfacing, electro surgery and dermabrasion, topical 5% imiquimod cream, and cryotherapy. Other medical therapeutic options are adalimumab (TNF blocker) and aspirin (inhibitor of NFkB).[4-7] Here we present this rare tumour with typical clinical and histological characteristics in 4 members of a family.

Case report

A 17 years old girl presented with 14 years history of multiple, pearly to skin colored, painless, non-pruritic nodular facial lesions which were cosmetically unacceptable to her. There was no history of drug reaction or trauma. She had no associated areas of hypopigmentation, neither was there any occular nor skeletal problem. Her paternal grandmother, uncle and brother had similar lesions. She took treatment from local hakim, but no relief. The main findings were multiple facial papules and nodules distributed on the nose, forehead and upper lip **(Figure 1)**. With positive family history of similar lesions (grandmother, brother and uncle) **(Figure 2a,2b,2c)**. A clinical diagnosis of MFT was made, which was later confirmed by histopathological studies of skin biopsy. Due to cosmetic and patient's psychosocial concerns, she was treated with electrocautery and 50% TCA. Histologically, the lesions showed multiple masses of basaloid cells. Multiple horn cysts were also seen. There

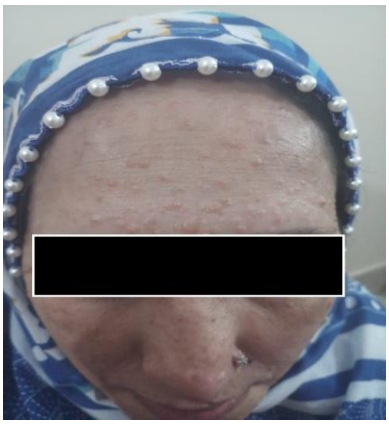

Figure 2a Patient's grandmother with multiple facial papules and nodules.

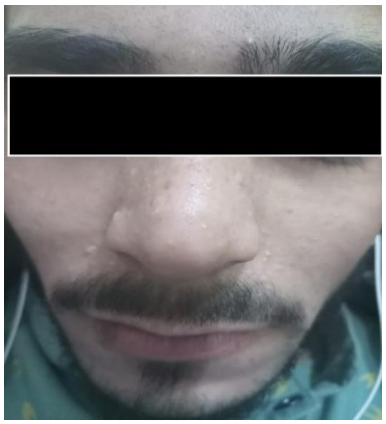

Figure 2b Patient's brother with multiple facial papules and nodules.

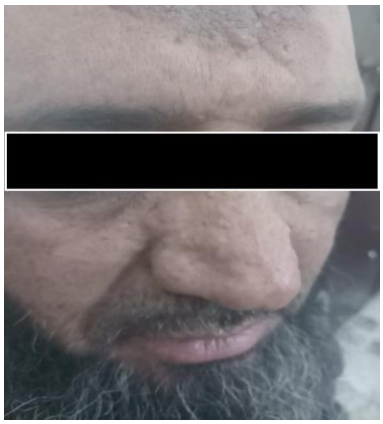

Figure 2c Patient's uncle with multiple facial papules and nodules.

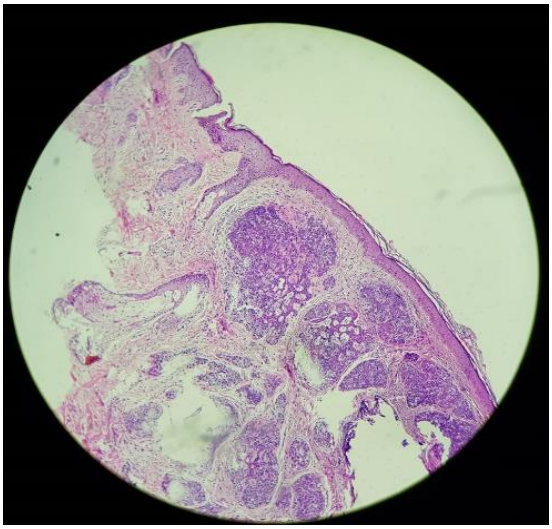

Figure 3a Photograph showing histology of TE.

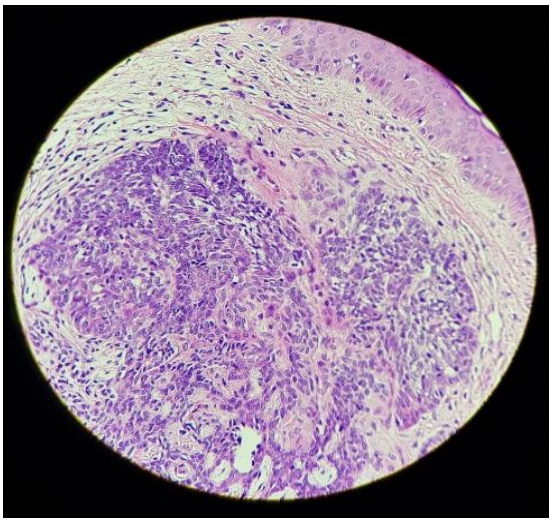

Figure 3b Histopathology showing multiple nests of basaloid cells embedded in connective tissue stroma.

was no evidence of granuloma or malignancy seen **(Figure 3a,3b)**. She was advised pulsed carbon dioxide laser but she opted for electrocautery with chemical cautery (50% TCA). She has undergone four sessions of electrocautery with chemical cautery. The lesions improved and the patient was satisfied.

Discussion

Patients with MFT most often present to the hospital for cosmetic reason and usually want treatment to improve their outlook. The lesions are usually disfiguring and cause psycho-social problem in patients, as seen in our patient who was not happy with her face even though the lesions were asymptomatic. In literature, there have been isolated case reports of this benign rare skin tumor but familial cases are quite rare and these cases are reported in two or three family members.[2,3,7] We could trace these lesions in four adult family members (two males and two females). One similar case of MFT in more than three family members had been published earlier in local literature.[2] In our case, we clinically suspected the lesions as trichoepithelioma, confirmed it histologically, diagnosed similar lesions in three other family members, counselled the patients and offered available treatment options.

The aim of highlighting this rare cutaneous tumour is to create awareness among dermatologists and plastic surgeons about its occurrence, tracing similar lesions in other family members, counselling the patients about its benign nature, addressing psychosocial concerns of the patients and offering appropriate therapeutic options including electrocautery, chemical cautery ablative lasers etc. Malignant change is possible as reports have shown that TE can undergo malignant transformation to BCC,[8] therefore such patients should also be followed up for any change in morphology of the lesions.

References

1. Yiltok S. Echejoh G. Mohammad A. Ituen A. Multiple Familial trichoepithelioma: A case Report and Review Of literature. *Nigerian J Clinical Practice*.2010;**13**:230-2.
2. Ejaz A. Raza N. Sohail M. Multiple familial trichoepitheliomas, *J Pak Assoc Dermatol*. 2005;**4**:207-209.
3. Bari AU, Rahman SB. Multiple familial trichoepithelioma: a rare cutaneous tumour. *J Coll Physicians Surg Pak*.2004;**14**:560-1.
4. Ali M. and Seyed J. Trichoepithelioma: A Rare but Crucial Dermatologic Issue. *World J Plast Surg*.2014;142-5.

5. Lefaki M. Theodosiou G. Delli F. Multiple Nonfamilial Unilateral Trichoepitheliomas: Report of a Case—Mini Review of the Literature. *Case Reports in Dermatological Medicine*.2019:6821854 (4 pages).
6. Sangwaiya A, Sharma J, Sharma S, Munghate A, Samal S, Sen R. Multiple Familial Trichoepithelioma with an Adjacent Basal Cell Carcinoma, Transformation or Collision- A Case Report and Review of Literature. *Indian J Dermatol*.2015;**60**;280–3.
7. Crotty K, Dutta B, Hogan P. Multiple trichoepitheliomas in a mother and daughter. *Austral J Dermatol*.2003;**44**;270-2.
8. Ayhan M, Adanali G, Senen D, Gorgu M, Erdogan B. Rarely seen cutaneous lesions in an elderly patient: Malignant transformation of multiple trichoepithelioma. *Ann Plast Surg*.2001;**47(1)**;98-99.