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Case Report

Multiple familial trichoepithelioma in mother and daughter

Bushra Bashir*, Zahida Rani, Rutaba Kiran*, Ijaz Hussain***

*Department of Dermatology, Unit I, King Edward Medical University/Mayo Hospital, Lahore
**Department of Dermatology, Khawaja Safdar Medical College/Allama Iqbal Memorial Hospital, Government Sardar Memorial Hospital, Sialkot

Abstract Trichoepithelioma is a benign adnexal tumour arising from hair matrix cells. It can be sporadic, familial or a component of other genetic diseases. We report a 40-year-old female who presented with disseminated papulonodular lesions on the face only which were confirmed histopathologically as trichoepithelioma. Her daughter also had trichoepithelioma in the nasolabial folds. They had no other systemic features and were concerned the cosmetic disfigurement imparted by the disease.

Key words
Familial trichoepithelioma.

Introduction

Multiple familial trichoepithelioma is a relatively infrequent familial disease. Because of its early onset, more expression in females and involvement of mainly exposed parts, it presents as a major cosmetic, social and psychological impairment in females. Although it is an autosomal dominant disorder but has less penetrance in males. We describe a case and her daughter who presented with multiple skin-coloured facial papules and nodules that caused a severe cosmetic disfigurement. Histopathology confirmed the diagnosis of trichoepitheliomas.

Case Report

A 40-year-old female patient presented to the dermatology department, Unit 1, Mayo Hospital, Lahore with a history of multiple skin-coloured papulonodular lesions on her entire face. These lesions started to appear nearly at 15 years of age and gradually increased in size and number till they involved the whole face causing severe disfigurement **(Figure 1)**. Similar lesions had started appearing in her daughter at the age of 12 years **(Figure 2)**. The patient denied a family history of any other cutaneous disorder or neoplasms. The patient had been otherwise healthy without any other medical problem, medication or systemic complaints.

Cutaneous examination showed abundant skin-coloured, dome-shaped papules and nodules mainly on the face, nose, cheeks, ears and forehead. The size varied from 1mm to 15mm in diameter, firm in consistency and without signs of inflammation (**Figure 1**). The scalp, neck, trunk, buttocks and extremities were unaffected. Skin appendages were normal. Examination of her daughter's face showed multiple skin-coloured 3-5mm papules in the nasolabial folds and adjacent areas of upper lip (**Figure 2**). Rest of her body was normal.

A skin biopsy revealed collection of basaloid cells with peripheral palisading along with hair follicles in between them. Horn cysts were seen. No retraction artefact was seen **(Figure 3)**.

Address for correspondence
Dr. Bushra Bashir, Assistant Professor,
Dermatology Department, King Edward Medical University/Mayo Hospital, Lahore
Email: bushra.b27@gmail.com

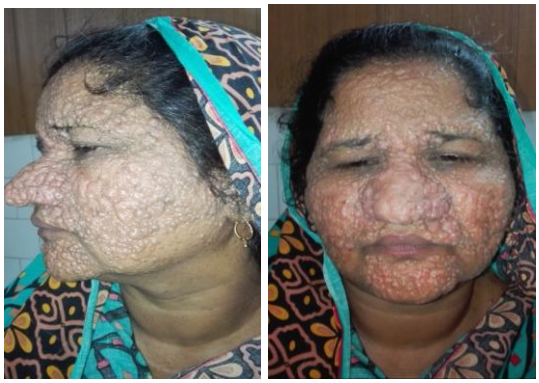

Figure 1 Innumerable skin-coloured, firm papulonodules on the whole face.

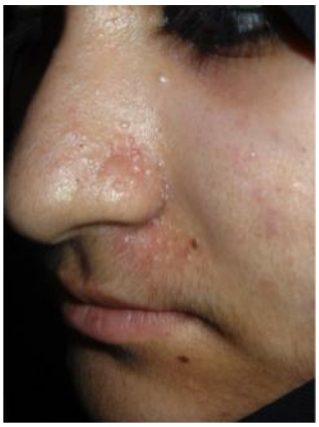

Figure 2 Skin-coloured, firm papules in the nasolabial fold extending to upper lip and left side of nose (daughter of the patient).

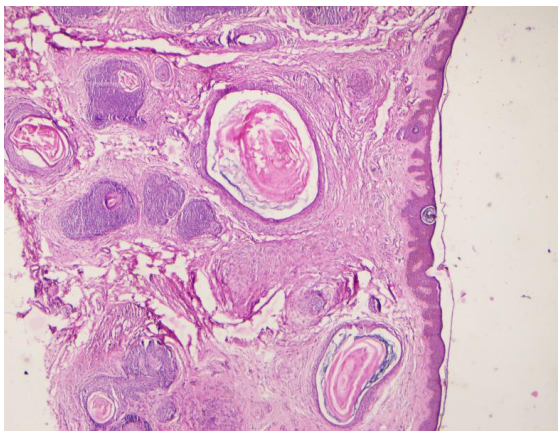

Figure 3 (a) Histopathology shows lobules of small dark basaloid cells with a degree of peripheral palisading surrounding a central area of eosinophilic amorphous material. A fibrous cellular stroma is seen around the cellular lobules. There is absence of retraction artefact.

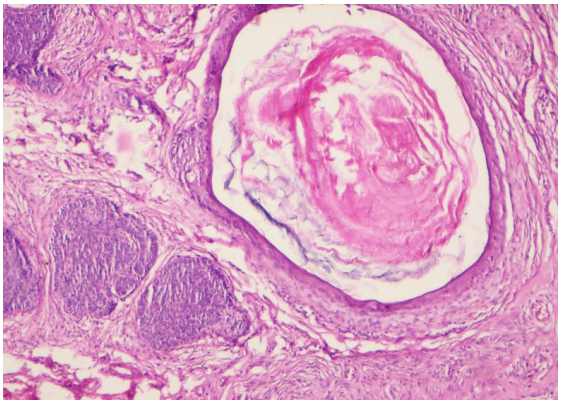

Figure 3 (b) Close-up magnified view of histopathology.

On the basis of clinical features, family history and histopathology, diagnosis of familial trichoepithelioma was made. Electrofulguration was advised for cosmetic treatment. Genetic analysis could not be performed due to unavailability.

Discussion

Multiple familial trichoepithelioma (MFT) is also called as epithelioma adenoids cysticum and Brooke-Fordyce trichoepitheliomas.[1]

Brooke-Spiegler syndrome, familial cylindromatosis and multiple familial trichoepithelioma are allelic and are considered to represent phenotypic spectrum of a single disease entity.

The gene involved in the familial form is located on band 9p21. Other cases are caused by mutations of the cylindromatosis oncogene (CYLD) which maps to 16/q12-q13.[2] This is an autosomal dominant disorder with less penetrance in men so mainly expressed in females. Significant number of Merkel cells within the tumor nests and CD34 positive dendrocytes show that it is derived from hair structures particularly the hair bulge.[1,2]

Hereditary forms present in younger individuals. The primary lesions are firm, skin-coloured papules and nodules, ranging in size from 2-8 mm. Some lesions may coalesce to form large nodules and tumours. Predominant sites are forehead, nasolabial

folds. 50% of the lesions occur on face and scalp. Lesions are usually bilateral.[1,3] Atypical forms involving one side, dermatomal and large plaque types have been reported. These lesions rarely ulcerate. These lesions enlarge slowly and then become stable at sometime. Three types of trichoepithelioma are known; solitary, multiple and the desmoplastic variety. They can only cause cosmetic disfigurement but occasionally basal cell carcinoma can occur in association with them.[4,5]

Trichoepithelioma should clinically be differentiated from basal cell carcinoma, trichilemmoma, trichofolliculoma, syringoma and colloid milium. Histopathological differential diagnoses are basal cell carcinoma, microcystic adnexal carcinoma, trichoadenoma and basaloid follicular hamartoma.[1,4,6,7]

Trichoepithelioma may be associated with other cutaneous tumours and may be a part of Rombo syndrome and Bazex syndrome.[1]

Histopathology typically shows horn cysts, tumor islands composed of basophilic cells of basaloid appearance arranged in peripheral palisading pattern.[4,5]

Treatment is mainly for cosmetic disfigurement. Various treatment modalities include surgical excision, chemical cauterization, laser resurfacing, electrosurgery and dermabrasion. 5% imiquimod cream applied topically has been useful. Other medical therapeutic options are adalimumab (TNF blocker) and aspirin (inhibitor of NF-kB).[1,3,4,7,8]

Our case represents familial hereditary trichoepithelioma, mother and daughter being affected. There was no family history of disease in the patient which may indicate a *de novo* mutation. However, genetic studies could not be done to detect the nature of mutation in our patient. Our patient's main concern was cosmetic ugliness.

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