Multiple familial trichoepithelioma: A case report

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Abstract
Trichoepithelioma are benign epidermal appendageal tumour with follicular differentiation. Tumor has been categorized into solitary, multiple and desmoplastic types. The multiple familial trichoepithelioma is autosomal dominant inheritance and rarely seen.

Key words: Trichoepithelioma, tumor, inheritance.

Introduction
The multiple familial trichoepithelioma is a skin tumour of autosomal dominant inheritance. We report family with multiple trichoepithelioma. Tumor causes only cosmetic disfigurement to the patients but occasionally basal cell carcinoma (BCC) can develop in association with trichoepithelioma.

Case history
A 19 year old female student attended in our clinic with asymptomatic multiple rounded skin coloured, firm, papulonodular lesions over face since 2 years of age. Multiple skin lesions first appeared at nose, gradually increased in number and size to involved nasolabial fold and adjacent area of cheek bilateral (figure 1). Her younger sister, who is 15 years old, also had similar lesions at nose and started at the age of 5 years (figure 2). Her father has multiple lesions of similar characteristics at face since the age of 6 years (figure 3). No other family members had similar lesions. There is no history of consanguinity in family.

Examination of lesion revealed multiple skin colored, firm papulonodular lesions of varying size at nose, nasolabial folds and adjacent skin at cheeks bilaterally. No telangiectasia and ulceration of lesions were observed at any time. General physical examination did not reveal any abnormality. She had normal intelligentsia. Routine lab investigation including x-ray skull was normal.

Skin biopsy from the lesions showed multiple horn cysts and tumor island composed of basophilic cells, which were normal in size separated by fibrous stroma and no cellular atypia and mitosis seen (figure 4). Diagnosis was consistent with trichoepithelioma.
Figure-1. Patient- Multiple skin colour firm papulonodular lesions of varying size at nose, nasolabial fold and adjacent area of cheek bilateral.

Figure-2. Father- Similar lesions at nose and nasolabial fold bilateral.

Figure-3. Younger sister- Similar lesion at nose

Figure-4. Skin biopsy showing multiple cornified cysts.
Recent studies reported a novel missense mutation in the CYLD gene (cylindromatosis oncogene). The gene associated with the familial type of trichoepithelioma links to the short arm of chromosome 9 and several tumor suppressor genes (ie, p16, p15, and the gene for the basal cell nevus syndrome) are encoded in this region. The gene for the development of familial trichoepithelioma also encodes for a tumor suppressor therefore if altered, cellular proliferation may be up-regulated because of a poorly functioning or absent tumor suppression.

Brooke–Spiegler syndrome (BSS, familial cylindromatosis or turban tumor syndrome) is an inherited disease characterized by neoplasms of the skin appendages such as cylindroma, trichoepithelioma, and spiradenoma. The disease has been mapped to 16q12-13, and mutations in the CYLD gene have been identified in families with this disorder. Of interest, multiple familial trichoepithelioma (MFT) has been described as a distinct disorder characterized by the familial occurrence of trichoepithelioma and has been mapped to 9p21. However, to date a candidate gene has not been identified. In this report, describe a four-generation family with BSS presenting predominantly with trichoepithelioma (resembling MFT phenotype). These findings exemplify clinical heterogeneity within BSS and mutations in CYLD are implicated in this disease. Although not conclusive, these findings suggest that BSS and MFT may represent a single entity.

Trichoepithelioma should be differentiated clinically with basal cell carcinoma, colloid milium, syringoma, trichilemmoma, trichofolliculoma. Multiple trichoepithelioma are benign and causes only cosmetic disfigurement to the patients but occasionally basal cell carcinoma (BCC) can develop in association with trichoepithelioma. Clinically, ulceration,
inflammation, necrosis, suggest a diagnosis of BCC and histologically, primitive hair structures, cornified cysts, cribriform pattern and stromal fibrosis favour a diagnosis of trichoepitheliom where as presence of mucine, stromal edema and retraction or claft around the basaloid islands suggest diagnosis of BCC.\textsuperscript{10}

Furthermore, focal positive CD34 staining of fibroblastic stroma has been reported in skin sections from trichoepithelioma and such staining is not seen in basal cell carcinoma.\textsuperscript{11}

**Treatment**

Solitary lesions can be excised. In the case of multiple tumors, this surgical approach may not be feasible. Split-thickness skin grafting, dermabrasion, and laser surgery have been proposed, but the results of these procedures vary.\textsuperscript{12,13}

Recurrence of solitary trichoepithelioma is uncommon. When the multiple facial lesions are surgically flattened by dermabrasion or laser therapy, they tend to regrow into elevated papules or nodules. This regrowth may occur rapidly within months, or it may take several years. Some patients find a prolonged cosmetic improvement and worthwhile even if repeated procedures are necessary. However, partial response for trichoepithelioma to 5% imiquimod cream has been reported.

**References**
