# Case 27

Facial papules running in a family

Tueboon Sriphojanart, MD.
Thanya Techapichetvanich, MD.

**Patient:** A 18-year-old Thai female from Nakornpratom

**Chief complaint:** Facial papules for 8 years

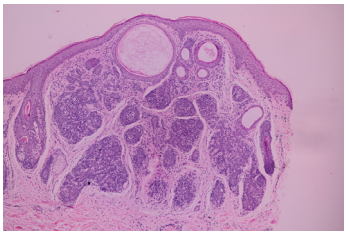
**Present illness:**

At the age of 10, the patient developed multiple, small, shiny, skin-colored, papules on her face, predominantly around the nose. These asymptomatic papules have gradually increased in size, as well as in number. She denied other skin lesions, or other systemic symptoms.

**Family history:** Her mother (individual I.2 in Figure 1) who deceased several years ago and her elder sister (individual II.1 in Figure 1) also have the similar skin lesions.

**Skin examination:**

- Multiple, small, shiny, skin- colored, papules on nose and both cheeks



**Physical examination:**

VS: BT $37.0^0$ c, BP 110/70 mmHg, HR 80/min, PR 80/min, RR 12/min
HEENT: Not pale conjunctivae, no icteric sclera, no oral ulcer
Heart: Normal S1 S2, no murmur
Lung: Normal breath sound
Abdomen: Soft, no hepatosplenomegaly
Neurological system: Intact
Musculoskeletal system: Intact

**Histopathology:** (S14-16693, face)

There are multiple cribiform aggregates of basaloid cells, some of which tend to from follicular structure within the fibrous stroma.

**Diagnosis:** **Familial multiple trichoepithelioma**
**Treatment:** Patient education and carbon dioxide laser resurfacing

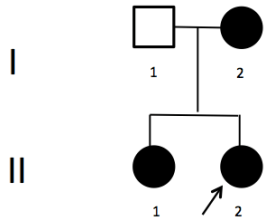



**Figure 1** Family pedigree of our patient.

## Discussion:

Multiple familial trichoepithelioma (MFT) is an autosomal dominant skin disease characterized by the presence of many small benign tumors with pilar differentiation, trichoepithelioma (TE) predominantly on the face.[1] Mutations in the CYLD gene have been identified as the cause of MFT.[2-3]

MFT is characterized by the development of multiple, firm, small, skin-coloured, shiny papules that originate from hair follicles. The papules most commonly occur between the first and second decade of life, predominantly on the face, nose, forehead and upper lip.

Multiple TEs can also associated with systemic lupus erythematosus, myasthenia gravis and Rombo syndrome, which characterized by atrophoderma, milia, hypotrichosis, basal cell carcinomas, and peripheral vasodilatation.

Histopathology of TEs present with sharply circumscribed, symmetric, dome shaped lesions composed of monomorphic basaloid cells aggregation in the upper dermis surrounded by abundant fibrous stroma with stromal- stromal retraction. The most common pattern is cribriform, however, nodular, and retiform patterns have also been observed. Trichoepitheliomas also exhibit periphral palisading and papillary mesenchymal bodies.

CYLD was originally identified as the human familial cylindromatosis tumour-suppressor gene. It is located on chromosome 16q12–13 and encodes the protein CYLD comprising 956 amino acids. CYLD is a deubiquitinating enzyme recently implicated in modulation of the nuclear factor-kB pathway and c-Jun N-terminal kinase (JNK) pathways, a crucial mediator of immune responses and inflammation. Skin appendages require NF-kB activity for their development. Impairment of apoptosis resulting from mutations in CYLD, which missense mutations lead to amino acid substitutions within the ubiquitin- specific protease (USP) domain of the CYLD protein[4-5], might deregulate proliferation in the developing skin appendages in familial cylindromatosis, Brooke- Spiegler syndrome as well as trichoepithelimatosis.

MFT is benign disease with rare report of malignant transformation.[6] MFT can be cosmetically disabling which affected patients may desire some type of intervention. Ablative approaches, including cryotherapy, electrosurgical destruction, and carbon dioxide laser resurfacing, have been employed with some success.[7-8]

## References: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8. Shaffelburg M, Miller R. Treatment of multiple trichoepithelioma with electrosurgery. Dermatol Surg. 1998; 24: 1154–6.