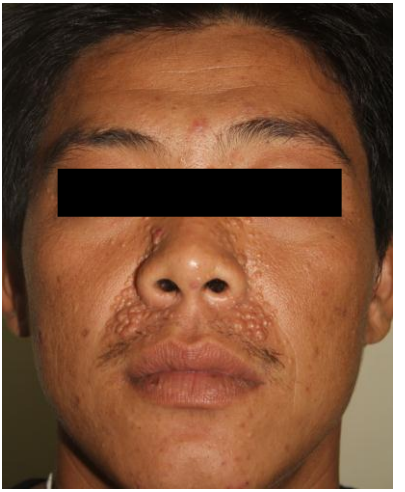
Case 9

A 29-year-old Thai male from Suphanburi.

Chief complaint: Multiple facial skin-colored papules for 14 years.



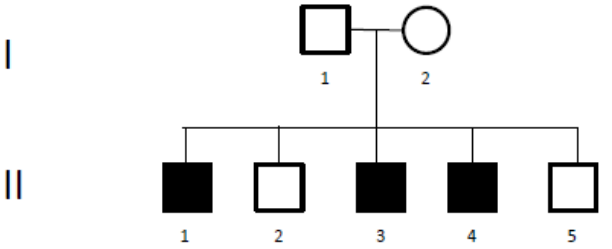
Present illness: The patient has developed multiple discrete asymptomatic skin-colored shiny papules on central face for 14 years. The lesions have gradually increased in number and size.

Past history

He has no underlying disease and history of seizure.

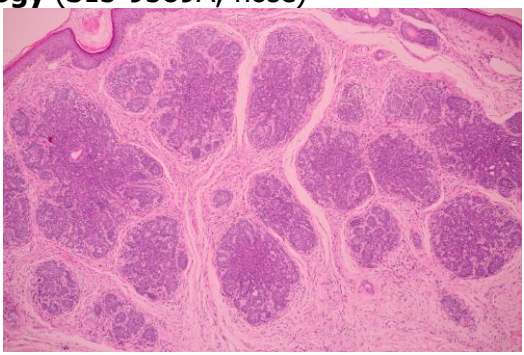
Family history

He is the third among five brothers. The oldest brother and the fourth brother also have similar lesions on their faces.



Skin examination
Multiple asymptomatic skin-colored shinny papules, 2-4 millimeters in diameter on nose and central face.

Histopathology (S13-9389A, nose)



There is well circumscribed tumor consisting of multiple cribifrom aggregations of basophilic cells within fibrous stroma. The tumor aggregates showing palisading nuclei, some of which tend to form follicular structure.

Diagnosis: Multiple familial trichoepitheliomas
Treatment: Carbon dioxide laser removal

Presenter: Sirima Sawatwarakul, M.D.
Consultant: Pranee Svetvilas, M.D.

Discussion

Trichoepithelioma is a rare benign tumor of the pilosebaceous follicles which appears predominantly in childhood and adolescence. Most lesions occur on face and also can occur on scalp.

Three distinctive patterns were reported including solitary, multiple and desmoplastic forms.

Solitary trichoepithelioma occurs mainly as small (5-8 millimeters) dome shape skin-colored papules on face especially on nose, upper lip, and cheeks.

Multiple familial trichoepitheliomas (MFT) is an autosomal dominant disorder characterized by groups of many small benign skin-colored papules predominantly on central face, scalp and upper trunk. They are usually developed during childhood.

While desmpoplastic form usually presents as a solitary, firm, skin-colored to white-gray, sclerotic annular plaque with central depression on cheek and angle of lip. It is rare to have multiple lesions. Desmoplastic trichoepithelioma can be differentiated from MFT by number and characteristic of lesions, family history of similar lesions, and histopathology that shows narrow strand of basalloid tumor cells, desmoplastic stroma, keratinous cysts, granuloma and calcification[1].

In MFT, the first locus was mapped on chromosome 9p21[2]. However recent studies show that mutations of the cylindromatosis tumor suppressor gene (CYLD) on chromosome 16q12-q13 have been identified as cause of MFT.

Loss of deubiquitinating activity of CYLD gene is considered as cause of tumor genesis[3]. Since CYLD gene has tumor suppressor properties and influences on cell proliferation, this gene has been shown to negatively regulate tumor necrosis factor alpha (TNF-α)-induced activation of nuclear factor kappa beta (NF-κB) and eventually resulting in tumor genesis[3].

Phenotypes of MFT that mutation in CYLD gene are indistinguishable from phenotypes assigned to chromosome gp21[4].

Mutation in CYLD gene has also been linked to Brooke-spiegler syndrome (BSS) and familial cylindromatosis (FC), therefore MFT are considered to be a phenotypic variant of BSS and FC.

Multiple trichoepithelioma were reported to be associated with Rombo's syndrome, SLE, myasthenia gravis and Bazex-Dupré-

Christol syndrome[5]. However in our patient, we don't find any associated conditions.

Although MFT has shown characteristic of benign tumor, malignant transformation to basal cell carcinoma rarely occurs.[6, 7]

MFT can be very disfiguring and lead to psychological distress.Treatment depends on number and extent of lesions. Solitary of few lesions can be excised but surgical excision may be difficult for numerous lesions. Laser surgery (CO2 laser and Erbium yag laser)[8-10], cryotherapy, electrosurgery, electrodesiccation, radiation and dermabrasion[11] had been reported. Recently there have been reported of treatment with topical imiquimod alone[12] and combined with topical tretinoin[13].

In our patient, we described a classic case of MFT present in young adult with autosomal dominant inheritance. We cannot identify any associated conditions and malignant transformation. He was treated with CO2 laser removal which plan for several session for cosmetic improvement.

References

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