Multiple Familial Trichoepithelioma: A Rare Clinical Entity

Dr Sreedevi P U¹, Dr Sreela L S², Dr Deepa Sujatha³, Dr Twinkle S Prasad⁴, Dr Philips Mathew⁵

¹Post graduate student, Department of Oral Medicine and Radiology, Government Dental College, Kottayam.
²Professor and HOD, Department of Oral Medicine and Radiology, Government Dental College, kottayam.
³Assistant Professor, Department of General Pathology, Government Medical College, Kottayam.
⁴Associate Professor, Department of Oral Medicine and Radiology, Government Dental College, Kottayam.
⁵Assistant Professor, Department of Oral Medicine and Radiology, Government Dental College, Kottayam.

Trichoepithelioma is an uncommon benign neoplasm of the pilosebaceous follicle that may present as non-hereditary solitary lesions or as multiple lesions that are often dominantly inherited. Multiple trichoepithelioma are inherited in an autosomal dominant manner and have been mapped to chromosome 9p21. They commonly present as multiple skin coloured papulo-nodular lesions in centro-facial distribution with an onset during early childhood or puberty. Here we present a case of Multiple Familial Trichoepithelioma in a 28 year old man who presented with multiple skin coloured facial papules and nodules with a history of similar lesion in other family members.
INTRODUCTION:
Trichoblastoma is a benign adnexal neoplasm that differentiates toward the trichoblast.¹ There are five types of trichoblastomas i.e. nodular, retiform, cribriform, racemiform, and columnar. Cribriform trichoblastoma is the most common pattern and is another name for trichoepithelioma.¹ Trichoepitheliomas present as solitary non-familial lesion or multiple lesions as a part of autosomal dominant inherited syndrome known as Multiple Familial Trichoepitheliomas (MFT) or Brook-Fordyce Disease.² MFT has been linked to genetic mutations in CYLD gene on chromosome16q12-13 and chromosome 9p.¹² It is an extremely rare condition with strong predilection for females. They commonly appear as multiple, skin coloured to pink, firm, papulonodular lesions which are mainly located at central part of the face during early childhood or puberty. Individual lesions reach a limiting size but the number may increase over the years producing significant cosmetic disfigurement. Continued growth and ulceration can raise a suspicion of change to basal cell carcinoma. Here, we describe a case of MFT in a 28 year old man who presented with multiple skin coloured facial papules and nodules with a history of similar lesion in other family members.

CASE REPORT
A 28 year old male patient presented with multiple asymptomatic papulo-nodular lesions which started over central part of the face and gradually increased in size and number since age of 3 years. No other lesions were present on other parts of the body. His father, elder brother, father’s brother and grandfather had history of similar lesions. Lesions in father appeared at the age of about five years around his nose and now whole face got involved causing cosmetic disfigurement. Eye problems were noted in both of them. His elder brother presented with only few facial papules and had no other associated eye problems. We plotted a family pedigree which showed that 11 members in four successive generation were affected with this disease.

On examination, multiple, asymptomatic, skin coloured, firm, papulonodular lesions measuring from 2 to 8 mm with smooth surface were present on central part of face. Single, smooth surfaced, skin coloured dome shaped nodule of size 1*1 cm seen on left cheek.

Thus based on history and clinical findings provisional diagnosis of Multiple Familial Trichoepithelioma was considered. Brook spiegler syndrome and familial cylindromatosis were included in differential diagnosis.

Histopathological examination of punch biopsy sample showed islands of basaloid cells with peripheral palisading, pseudohorn cysts and hair follicles, thus confirming the diagnosis of trichoepithelioma. Based on clinical and histopathological findings, diagnosis of Multiple Familial Trichoepithelioma was made.

Family pedigree showing eleven members in four successive generations affected with disease.

Fig 1: Family pedigree showing eleven members in four successive generations affected with disease.
Fig 2: Centrofacial distribution of papulo-nodular lesion

Fig 3: Single, smooth surfaced, skin coloured dome shaped nodule of size 1*1 cm seen on left cheek.
Fig 4 Multiple Trichoepitheliomas over face in his father

Fig 5: Histopathology showing horn cysts and tumor islands of basaloid cells with peripheral palisading.
DISCUSSION
Trichoepithelioma is a benign cutaneous tumor that originates from hair follicles and can be presented as solitary or multiple, familial or non-familial. Multiple Familial Trichoepithelioma is a rare autosomal dominant inherited disorder with an annual incidence of 2.14-2.75 cases. In MFT individuals presents with multiple skin coloured centrofacial papules of 2-8mm especially in nasolabial folds and between the nose and lips, but may occur on the neck, scalp and trunk. These lesions typically begin to develop during childhood or adolescence. They will increase in number but reach a limiting size with age and whole face will got involve producing significant cosmetic disfigurement. Involvement of eyes, ears can affect the vision and hearing. Here the clinical findings and family history were typical of MFT. Both sexes can be equally affected but females show strong predilection because of lessened expressivity and penetrance in males. In contrast, here majority of family members affected are males. Literature reveals evidence of genetic heterogeneity within MFT. Early reports linked MFT to chromosome 9p21 but recent reports suggests of mutation in Cylindromatosis Tumor Suppressor (CYLD) gene, which maps to chromosome 16q12-q13. Associated with MFT, seventeen mutations of this gene have been described. CYLD protein act as tumor supressor gene so mutations within this results in tumorgenesis. As Brooke Spiegler Syndrome (BSS) is also associated to CYLD gene, MFT can likely be a phenotypic variant of BSS. BSS is an autosomal dominantly inherited syndrome characterised by multiple skin appendageal tumors such as trichoepitheliomas, cylindromas and spiroadenomas. BSS, Familial Cylindromatosis and MFT share overlapping clinical findings. Familial Cylindromatosis is characterised by cylindromas and MFT by trichoepitheliomas as the only tumor type. Since all these conditions linked to mutations in CYLD gene, histopathology plays an important role to distinguish between BSS, MFT and Familial Cylindromatosis. Multiple trichoepitheliomas also form a part of other rare syndromes like the Rombo syndrome, Basex syndrome and cowden syndrome. On histologic examination, trichoepithelioma appears to be well circumscribed tumor with horn cysts the most characteristic feature. Tumour islands composed of basophilic cells of basaloid appearance arranged in peripheral palisading pattern. In our case histopathology findings suggests of typical features of trichoepithelioma. In certain cases very few or absence of horn cysts were noted which makes

Fig 6: High power field of view showing basaloid cells with peripheral palisading.
difficulty in differentiation of trichoepithelioma from Basal Cell Carcinoma. Malignant transformation of these lesions to basal cell carcinoma is rare. Any suspicious lesion should call for excision and histological examination.

Various treatment modalities have been used for treatment of trichoepithelioma. A solitary lesion can be treated by surgical excision. The combination of carbon dioxide and erbium: yttrium aluminium garnet laser ablation have been advocated as a useful treatment. Topical imiquimod cream also had good results. Other treatments include cryosurgery, dermabrasion, photodynamic therapy and other medications. However in most of the cases no regress was seen. Our patient was undergoing carbon dioxide laser ablation from Department of Dermatology since 2 months and he was under follow up. Wait and watch policy along with assurance is a more advisable strategy, as most of the lesions of MFT regresses with passage of time.

CONCLUSION

MFT is relatively an uncommon inherited benign adnexal neoplasm that can be diagnosed by the centrofacial distribution pattern of papules and nodules, positive family history and by characteristic histopathological findings of horn cysts and tumor islands of basaloid cells. Even carcinomatous changes have been observed in these lesion, therefore close follow up of patient is recommended.

REFERENCES


How to cite this article:

Dr Sreedevi P U, Dr Sreela L S, Deepa Sujatha, Twinkle S Prasad Multiple Familial Trichoepithelioma: A Rare Clinical Entity Br J Pharm Med Res , Vol.03, Issue 05, Pg.1325-1330, September - October 2018. ISSN:2456-9836 Cross Ref DOI : https://doi.org/10.24942/bjpmr.2018.364

Source of Support: Nil Conflict of Interest: None declared
Your next submission with **British BioMedicine Publishers** will reach you the below assets

- Quality Editorial service
- Swift Peer Review
- E-prints Service
- Manuscript Podcast for convenient understanding
- Global attainment for your research
- Manuscript accessibility in different formats
  - (Pdf, E-pub, Full Text)
- Unceasing customer service

Track the below URL for one-step submission

http://www.britishbiomedicine.com/manuscript-submission.aspx