Multiple Familial Trichoepithelioma

Alternative Names
MFT
TEM
Epithelioma Adenoides Cysticum of Brooke
EAC
Epithelioma, Hereditary Multiple Benign Cystic Brooke syndrome
Epithelioma Adenoides Cysticum

WHO International Classification of Diseases
Neoplasms

OMIM Number
601606

Mode of Inheritance
Autosomal dominant

Gene Map Locus
9p21

Description
Trichoepithelioma is a benign skin tumor of the pilosebaceous follicle. It may occur either as nonhereditary solitary lesion or as multiple lesions (multiple trichoepitheliomas), which may be a new or autosomal dominant germline mutation. Trichoepithelioma usually appears in childhood or early adolescents and may involve a limited area of the body, most commonly on the face, or be widespread. Multiple hereditary trichoepithelioma was first described by Brooke in 1899 and was previously also known as Brooke syndrome and epithelioma adenoides cysticum. When occurring as the only cutaneous finding, it is called trichoepithelioma papulosum multiplex. Alternatively, it may be associated with other cutaneous findings such as cylindromas, spiradenomas, and milia, in a syndrome called Brooke-Spiegler. The differential diagnosis of multiple familial trichoepithelioma includes BCC, other appendageal tumors, syringoma and angiofibroma.

Treatment options include surgery, split-thickness skin grafting, dermabrasion cryotherapy, electrodessication, and carbon dioxide laser. All methods carry significant risk of side-effects, and most importantly scarring and unsatisfactory results.

Molecular Genetics
Genetic studies in families with multiple familial trichoepithelioma, two African American and one Caucasian, mapped the gene for multiple familial trichoepithelioma to chromosome 9p21 in the 4-cM region between IFN-alpha and D9S126. Several tumor suppressor genes have been mapped to this region, suggesting that the gene for multiple familial trichoepithelioma may be a tumor suppressor gene. It is also thought that multiple familial trichoepithelioma may be caused by more than two independent genes: one that occurs in isolation and is determined by a gene on 9p, and another due to impairment of the gene for cylindromatosis on chromosome 16. Recent research has firmly established that familial trichoepithelioma is due to germline mutations in the cylindromatosis gene.

Epidemiology in the Arab World

Saudi Arabia
Al Aboud et al. (2006) described multiple hereditary trichoepithelioma in a Saudi kindred, affecting 14 patients and spanning five generations. Clinical examinations were performed on the sibship, comprising of two sisters and a brother, in the 5th generation, and their mother. The mother, previously diagnosed with diabetes and epilepsy, presented with numerous skin-colored, smooth, round papules and nodules involving the face, scalp, and upper region of the back. Her son, a 27-year-old man, showed tiny papules symmetrically distributed on the nasolabial folds on the face on both sides. He was earlier diagnosed with epilepsy and was
withdrawn from school at early age due to mental deficiency. The eldest sister, 21-year old, presented with multiple papules involving the face, scalp, and shoulders, and no other systemic disease. The younger, 19-year old sister had multiple papules and nodules of the face, nodules around the right eye interfere with vision, and papules in the scalp coalesce to form multiple plaques of alopecia. A large exophytic nodule (5 cm) on her scalp was proven by histopathology to be trichoepithelioma. Al Aboud et al. (2006) noted that the kindred described represented one of the largest families affected with this disease to be reported. Al Aboud (2006) further remarked that it is unclear whether epilepsy and low intelligence are associations with trichoepithelioma or incidental findings.

References

Contributors
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