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Multiple familial trichoepithelioma

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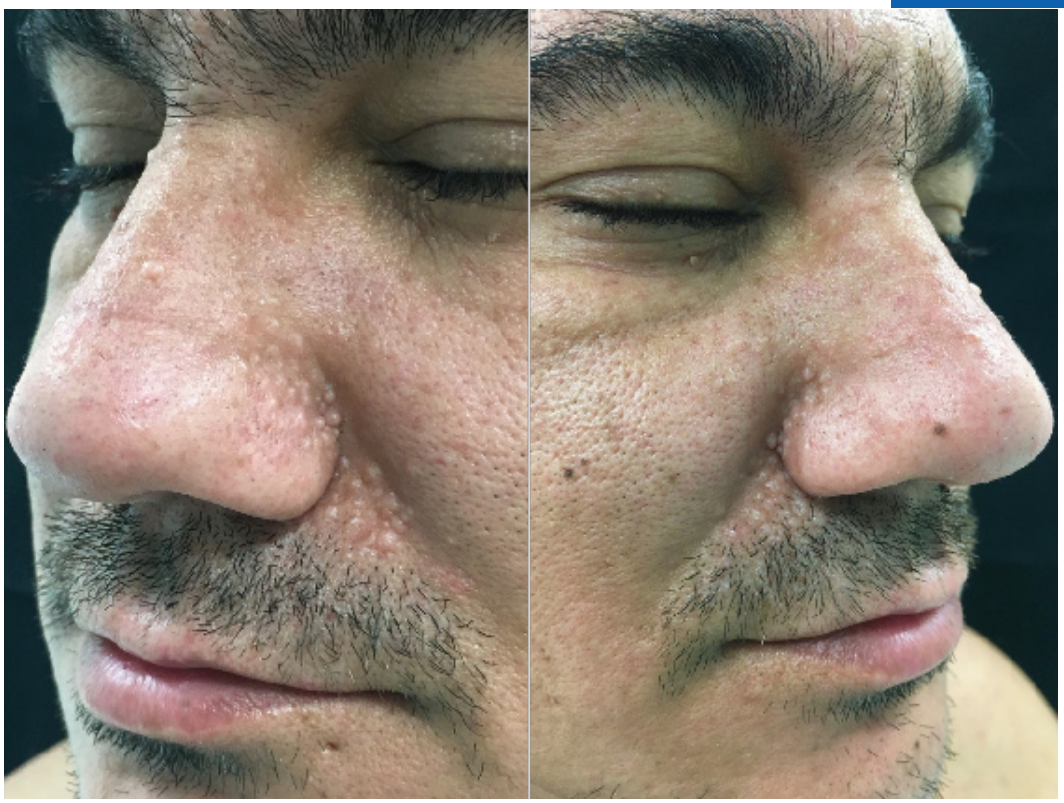
Description

A 33-year-old male presented to our dermatology department with multiple skin-colored dome-shaped papules and nodules located on the central area of the face: forehead, nose, nasolabial folds and perioral region (Figure 1A & 1B). Lesions were asymptomatic, slowly developing since childhood and increasing in number and size over the years. The patient reported his mother, sister, maternal grandmother and 3 maternal aunts had similar lesions. Excision biopsy and histopathologic examination revealed trichoepitheliomas. Genetic study showed a pathogenic heterozygotic mutation in exon 9 of the CYLD gene [c.1112C>A (p.Ser371*)], supporting the clinical diagnosis of Multiple Familial Trichoepithelioma (MFT). Surgical excision of the largest lesions and treatment with CO_2 laser, cryotherapy and electrosurgery were offered but the patient refused treatment. The patient and family were referred to the genetics department.

Trichoepitheliomas are rare, benign, poorly differentiated hamartomatous tumors originating from the pilosebaceous follicle. MFT is an uncommon autosomal-dominant genodermatosis characterized by the appearance of multiple skin-colored, symmetrical papules and nodules predominantly located on the central areas of the face – nasolabial folds, nose, forehead and upper lip – and occasionally on the scalp, neck and upper trunk. Recent studies have reported a mutation in the Cylindromatosis tumor suppressor gene (CYLD) located in the chromosome 16q12-q13 in roughly 40% of cases. MFT can be cosmetically and psychologically disabling and treatment options generally have unsatisfactory outcomes. The association of MFT with malignancy is rare, however, trichoepitheliomas can infrequently undergo malignant transformation to basal cell carcinoma. It is therefore important to recognize this genodermatosis so that patients can be kept under long-term observation and referred for genetic counselling.



Figures 1A & 1B: Skin-colored papules on the central area of the face

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