Multiple Familial Trichoepithelioma

Tricoepitelioma Múltiplo Familiar

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A 62-year-old woman presented with multiple skin-coloured papules and nodules on the scalp, face, intergluteal cleft which had developed slowly since childhood (Fig. 1). Her mother, three sisters and two daughters had similar lesions. Recently, she noticed a rapid enlargement of a 12 mm nodule on the right temporal area. Excision biopsy and histopathologic examination of a facial and sacral lesion revealed trichoepitheliomas (TE) (Fig. 2), whereas the temporal nodule was compatible with basal cell carcinoma (BCC). Genetic study showed a heterozygotic mutation in exon 9 of CYLD gene [c.1112C > A(p.Ser371*)], supporting the diagnosis of multiple familial trichoepithelioma (MFT).

MFT is an uncommon autosomal-dominant genodermatosis characterized by multiple TE on the face, scalp and, neck with rare reports on other anatomical locations. The association of MFT with malignancy is rare, however, TE can infrequently undergo malignant transformation to BCC. That is why these patients should be kept under long-term observation.

REFERENCES