Folliculocystic & Collagen Hamartoma of Tuberous Sclerosis Complex: A Case Report

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Introduction

Many people with tuberous sclerosis complex (TSC) have cutaneous manifestations: hypomelanotic macules (ashleaf macules)(97.2%), facial angiofibromas (74.5%), shagreen patch (plaque of collagenoma) (48%), periungual fibroma (Koenen's tumor)(15.1%), and fibrous cephalic plaque (18.9%). Folliculocystic and collagen hamartoma (FCCH) is an uncommon cutaneous manifestation of TSC presenting as a complex hamartoma with thick collagen deposition, concentric perifollicular fibrosis, and keratin-filled infundibular cysts. On physical exam, the lesions appear as "large, painless, infiltrated plaques studded with follicular comedo-like openings and cysts containing and draining a keratinous or purulent material". Since its initial characterization, 12 cases of FCCH have been reported.

Case Presentation

A 17-year-old male with a known history of TSC and multiple angiofibromas on the face presented to the dermatology clinic with a large, painless exophytic mass on his central forehead requesting excision due to cosmetic concern (Figure 1). Microscopic examination of the lesion demonstrated epidermal papillomatosis, cystically dilated hair follicles (Figure 2A), hyperplastic sebaceous lobules displaced to the lower portions of the dermis by the cystic expansion of the follicular infundibulum (Figure 2A), and increased vascularity and fibrocytes (Figure 2B). There was also an expansion of the subcutaneous collagenous tissue which greatly contributed to the nodularity of the lesion noted clinically (Figure 2C).

Three previous of excisions taken from similar nodular lesions on the forehead and scalp show similar histopathologic changes consistent with FCCH (Figure 3). Previous excision material also showed follicular cysts which had ruptured and an associated granulomatous inflammatory reaction with associated perifollicular fibrosis . The lesions were initially diagnosed as dissecting cellulitis, but on further review at the time of subsequent biopsies were found to meet the criteria for a diagnosis of FCCH.

Clinical Photos and Pathology

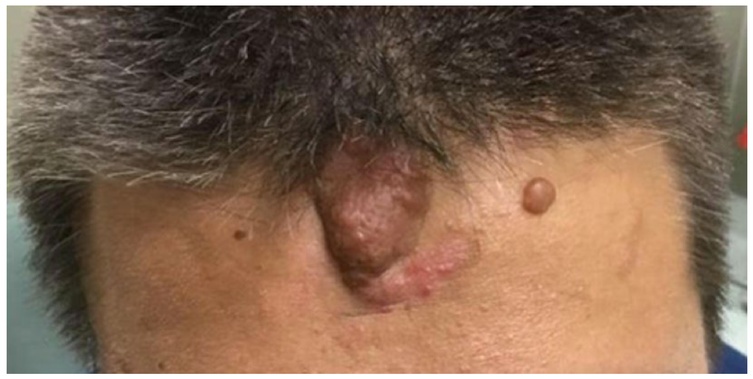

Figure 1. Large exophytic mass on the central forehead, with prominent comedones.

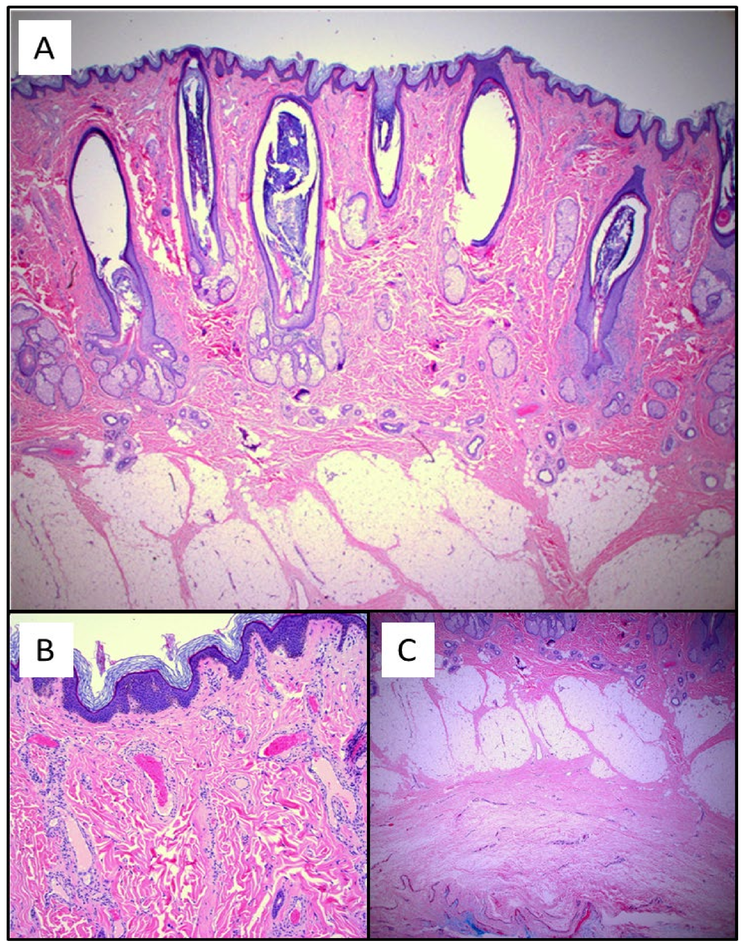

Figure 2. Photomicrograph of the exophytic forehead lesion stained with H&E demonstrating A) dilated, cystic hair follicles with hyperplastic sebaceous glands displaced to the lower portion of the dermis. B) Increased vascularity and fibrocytes. C) Expansion of subcutaneous collagen underlying normal adipose tissue.

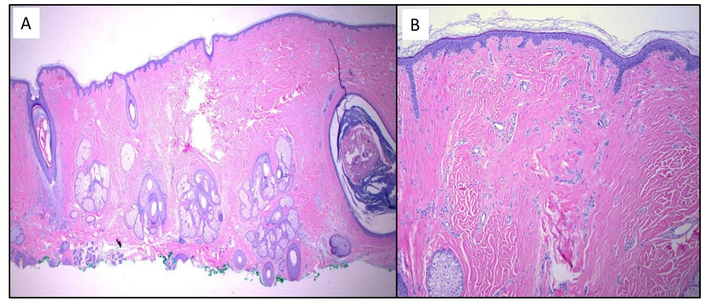

Figure 3. Tissue from nodular lesions previously excised from the patient's scalp and forehead stained with H&E demonstrating A) dilated, cystic hair follicles with hyperplastic sebaceous glands displaced to the inferior aspect of the dermis and B) abundant dermal collagen.

Discussion

FCCH is a recently described cutaneous manifestation of TSC with a distinct combination of histopathologic features. However, individually such features resemble other cutaneous lesions associated with TSC. The increased number of blood vessels and fibrocytes resemble angiofibroma. Accentuated collagen deposition is a feature of fibrous cephalic plaque and shagreen patch. Although not typically associated with TSC, it is worth noting that the papillomatous epidermis and prominent dermal sebaceous units seen in this case can be seen in nevus sebaceous. Until FCCH was formally described in 2011, there was no single entity characterized by these three histologic characteristics.

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