

Case Letter

Multiple Familial Trichodiscomas

To the Editor:

Trichodiscoma is a rare benign tumor first described by Pinkus et al¹ in 1974. It represents a hamartomatous growth that recapitulates the hair disk, fibrous sheath, or papilla; produces a dome-shaped papule; and is characterized by a well-circumscribed proliferation of loosely packed fibroblasts and blood vessels. Mucin accumulation and a hair follicle at the margin of the lesion usually are found.¹ It can occur either as a solitary papule or multiple tumors, with the face being the most commonly affected site. Trichodiscomas have been described in association with fibrofolliculomas and acrochordons configuring a dominantly inherited disease, also known as Birt-Hogg-Dubé syndrome.² One case was found in conjunction with colonic polyposis.³ Multiple trichodiscomas in the absence of other cutaneous or extracutaneous disorders rarely have been reported among members of the same family.⁴ We report a case of multiple familial trichodiscomas.

A 29-year-old man who was otherwise healthy presented with several firm papules on the face, both ear auricles, and elbow (Figure 1). They appeared as dome-shaped pink lesions measuring 2 to 4 mm in diameter. Ten years prior the lesions had presented on the right side of the zygoma and subsequently underwent progressive dissemination. Several punch biopsies displayed the same histologic characteristics. Following the diagnosis in this patient, his living family members were examined, and his maternal grandfather and mother demonstrated multiple similar lesions. His grandfather had approximately 200 papules on his face ranging from 0.5 to 5 mm in diameter that had developed during the last 10 years. His mother only had 10 elements that also were located on the face. Several 4-mm punch biopsies were performed in both family members, which revealed identical histologic features. No additional

cutaneous or extracutaneous lesions were detected in the 3 family members.

Light microscopy showed that the papules of the 3 patients were all characterized by a loose proliferation of fibroblasts that contained mucin deposits and blood vessels (Figure 2). Neither pleomorphism nor hyperchromasia were detected. No mitotic figures were found. Thick myelinated nerve fibers and occasional hair follicles were detected at the bottom and edge of the lesions, respectively.

Immunohistochemistry was performed on paraffin sections and revealed clear-cut CD34 positivity of



Figure 1. Trichodiscoma observed on the elbow.

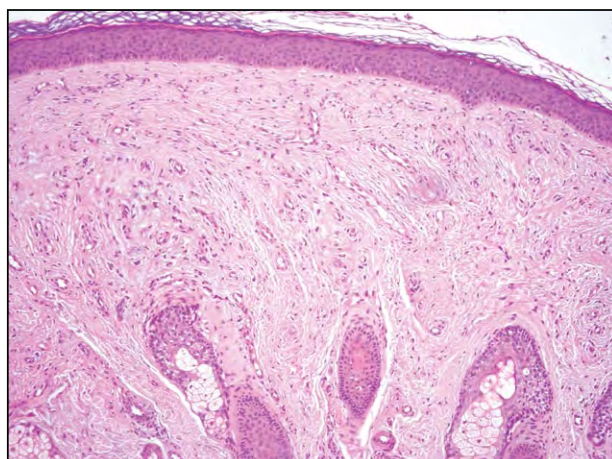


Figure 2. At low power, the lesion is characterized by a loose proliferation of fibroblasts and blood vessels (H&E, original magnification $\times 100$).

From Dermatology, Department of Specialised, Experimental and Diagnostic Medicine, University of Bologna, Italy.

The authors report no conflict of interest.

Correspondence: Alessandro Pileri, MD, Dermatology, Department of Specialised, Experimental and Diagnostic Medicine, University of Bologna, Via Massarenti 1, Bologna 40138, Italy (alessandropileri@hotmail.it).

fibroblasts and blood vessels as well as the substantial lack of Merkel cells in the overlying epidermis. A few nerve fibers at the inner borders of the tumors expressed S-100 protein. According to the morphological and immunohistochemical findings, a diagnosis of trichodiscoma was made in all 3 patients.

Neoplasms of the pilosebaceous unit can be classified into 2 groups: (1) of epithelial derivation (eg, trichofolliculoma, trichoepithelioma, and tricholemmoma) and (2) of mesenchymal origin (eg, trichodiscoma, perifollicular fibroma). Fibrofolliculoma has intermediate features between these 2 groups. It may be difficult to distinguish trichodiscoma and fibrofolliculoma.⁵ However, fibrofolliculoma demonstrates a central hair follicle with numerous anastomosing bands of follicular epithelium extending into the stroma, which is not observed in trichodiscoma.⁵ Additional differential diagnoses include focal dermal mucinosis, dermal myxomas, and neurofibromas with mucinous change. Unlike mucinous degenerative lesions, trichodiscoma has a characteristic architecture that is highlighted by CD34 staining. Neurofibromas extend more deeply into the dermis than trichodiscomas, and they contain mast cells and more nerve twigs that are clearly shown by S-100 protein positivity.⁵

The lesions described in our case met the morphologic and immunophenotypic criteria for the diagnosis of trichodiscoma, including the lack of Merkel cells in the overlying epidermis. We observed lesions that presented as a dominantly inherited disease in 3 members of the same family. Notably, unlike other

reports in the literature,² they were not associated with fibrofolliculomas and acrochordons (thus configuring the Birt-Hogg-Dubé syndrome) or colonic polyposis,³ which strengthens the concept that multiple trichodiscomas can be hereditary.⁴ Clinicians should be aware of this possibility and carefully gather the family anamnesis for patients showing multiple trichodiscomas to conceivably examine their relatives. The occurrence of a hereditary disorder suggests further evaluation of the family history for possible implications on the future progeny.

Iria Neri, MD

Carmine D'Acunto, MD

Alessandro Pileri, MD

Annalisa Patrizi, MD

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