Multiple familial trichoepithelioma: confirmation via dermoscopy

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Trichoepitheliomas are uncommon benign adnexal neoplasms that originate from the hair follicles. Multiple familial trichoepitheliomas constitute an autosomal dominant disease characterized by the appearance of multiple flesh-colored, symmetrical papules, tumors and/or nodules in the central face and occasionally on the scalp. Although clinical diagnosis is usually straightforward in light of the family history and naked-eye examination, dermoscopy may aid in its confirmation. Dermoscopy of each papule revealed in-focus arborizing vessels, multiple milia-like cysts and rosettes amidst a whitish background.

In a patient with multiple facial papules revealing a dermoscopic appearance described above, the diagnosis of sporadic or familial multiple trichoepithelioma should be considered.

Introduction

Trichoepitheliomas are uncommon, benign, adnexal neoplasms that originate from the hair follicles. They usually present as solitary lesions but in the familial setting they appear as multiple lesions. Multiple familial trichoepithelioma constitute an autosomal dominant disease characterized by the appearance of multiple flesh-colored, symmetrical papules, tumors and/or nodules located in the central face and occasionally on the scalp [1].

Herein we present an interesting case of a female with multiple facial papules easily diagnosed as trichoepitheliomases with the use of dermoscopy.

Case report

A 29-year-old otherwise healthy female presented to our clinic for evaluation of multiple firm, flesh-colored, dome-shaped papules distributed primarily along the nasolabial folds and forehead. She first noticed their appearance during
monly present as solitary papules, but in the familial setting they appear in clusters characteristically involving the central face and/or the scalp [2]. They present with variable size from small papules that are of minor cosmetic relevance to multiple tumors that can lead to functional impairment such as visual obstruction. The incidence of multiple familial trichoepithelioma (OMIM #601606) in the US has been reported to be between 2.14—2.7 cases per year, with a female predilection [2,3]. They frequently first appear during childhood or adolescence.

Discussion

Trichoepitheliomas are rare benign hamartomatous tumors originating from the pilosebaceous follicle. They most commonly present as solitary papules, but in the familial setting they appear in clusters characteristically involving the central face and/or the scalp [2]. They present with variable size from small papules that are of minor cosmetic relevance to multiple tumors that can lead to functional impairment such as visual obstruction. The incidence of multiple familial trichoepithelioma (OMIM #601606) in the US has been reported to be between 2.14—2.7 cases per year, with a female predilection [2,3]. They frequently first appear during childhood or adolescence.
When evaluating a solitary papule with the aforementioned dermoscopic findings, a biopsy may be necessary for definitive diagnosis. If a biopsy is contemplated, then it is important to underscore that small partial biopsies may lead the pathologist to misinterpret the specimen as basal cell carcinomas [12]. Histopathology findings suggesting the diagnosis of trichoepithelioma over basal cell carcinoma include the fibrocytic loss of the stroma, aggregations of cells with smooth borders, the presence of granulomatous inflammation, monomorphic nuclei and papillary mesenchymal bodies [1].

Finally, treatment options for multiple trichoepithelioma include destructive/ablative techniques such as cryotherapy, dermabrasion, electrodissection and radiation therapy, all with variable to poor outcomes [2]. Interestingly, our patient chose not to undergo any treatment, which is also a valid option.

**Conclusion**

A patient evaluated with multiple facial papules that on dermoscopy reveal in-focus arborizing vessels, multiple milia-like cysts and rosettes amidst a whitish background should lead the clinician to consider the diagnosis of sporadic or familial multiple trichoepithelioma.

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