Dermatology Practical & Conceptual

An Atypical Presentation of BAP1-Inactivated Melanocytic Tumor

Sara Pilar Herrero-Ruiz[1], Helena Álvarez-Garrido[1], Anastasia Alejandra Garrido-Ríos[1], Laura Fernández de la Fuente[1], Radia Khedaoui[2], Jesus Borbujo[1]

1 Department of Dermatology, Hospital Universitario de Fuenlabrada, Madrid, Spain
2 Department of Pathology, Hospital Universitario de Fuenlabrada, Madrid, Spain

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Corresponding author: Sara Pilar Herrero-Ruiz (MD), 2 Molino Street, Fuenlabrada, Madrid, 28942 Spain. E-mail: sarahr-6@hotmail.com

Introduction

BAP1-inactivated melanocytic tumor (BIMT) is considered a type of intermediate melanocytic tumor [1]. Clinically BIMTs are described as a unique papule from 2 to 10 mm in diameter. The size and number of BIMTs in our patient are not consistent with these previous descriptions.

Case Presentation

A 47-year-old woman presented with a lesion on the scalp that had been stable since childhood, however during the last 6 months it had experienced a progressive enlargement. The patient had family history of different malignancies. Her mother and aunt had had non-melanoma skin cancer, father had had leukemia, several uncles had had lung and breast cancer, and her grandparents had had pancreatic and lung cancer.

Physical examination showed a 2-cm pink pedunculated hyperkeratotic lesion located in the interparietal region, with multiple surrounding smaller erythematous papules (Figure 1, A and D). Dermoscopy showed multiple telangiectasias and whitish scales on a pink background (Figure 1, B and C).

The largest lesion was excised, and histopathology revealed that the dermis was expanded by a cellular substitution arranged in nests and streams (Figure 2A). Some cells had a nevic appearance and were epithelioid in morphology (Figure 2B), occasionally pigmented, and extended deep surrounding the adnexal structures, with no tendency to maturation. They presented well-defined limits, a wide eosinophilic cytoplasm, prominent nucleoli and pseudoinclusions. It also showed a focus with nests of a conventional nevus (Figure 2C).

Immunohistochemistry obtained a positive result for melan-A in the entire sample (Figure 2D). BAP1 expression was lost except in the conventional-appearing nevus component (Figure 2E), so the final diagnosis was a BIMT.

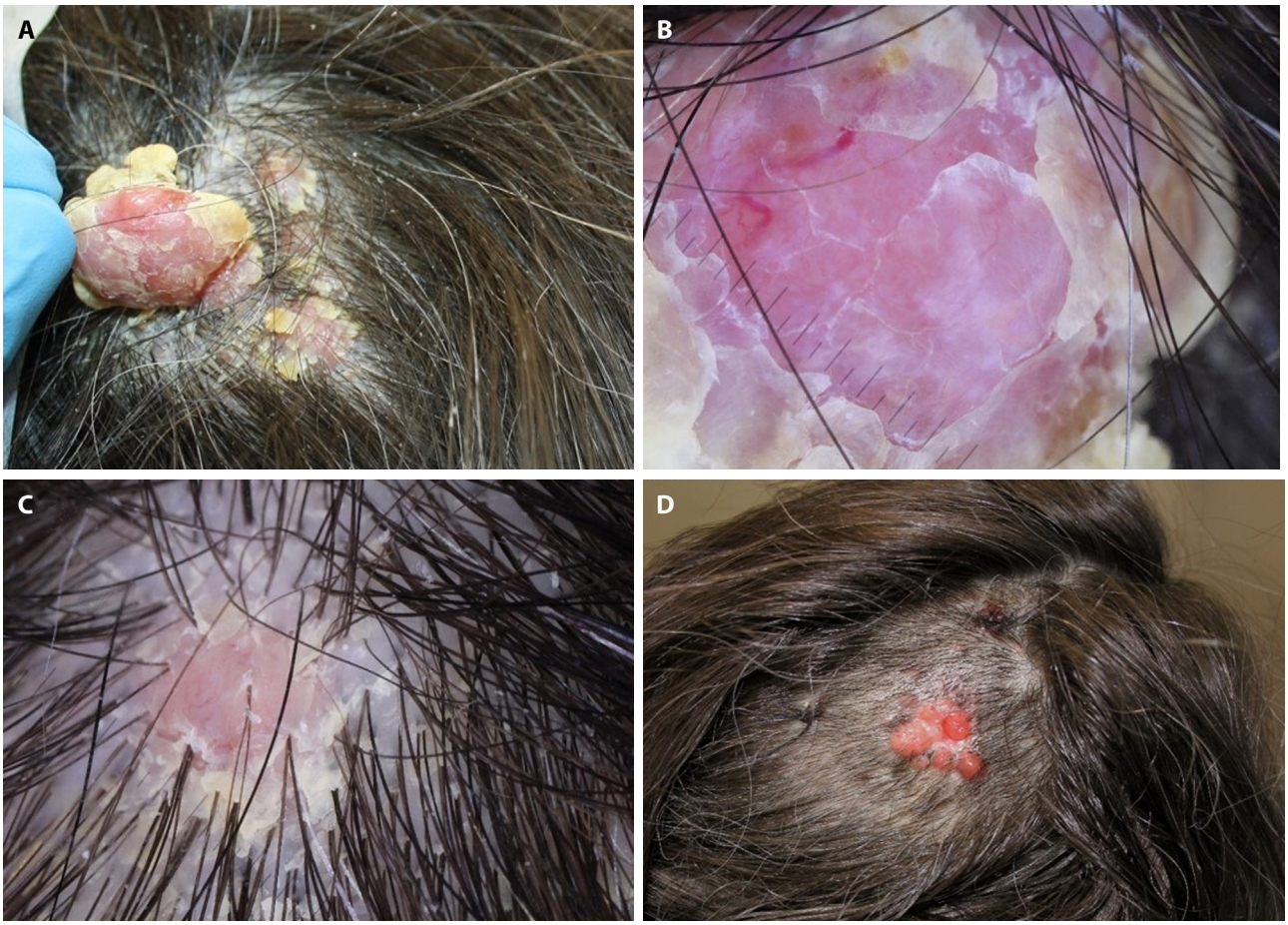

Figure 1. (A) Erythematous pedunculated papule with yellow and whitish scales on its surface and multiple small similar lesions on the base. (B) Dermoscopy of the biggest lesion showed thick central telangiectasia with several thinner vessels in the rest of the lesion, on a pink background with whitish scales. (C) The smaller papules had similar dermoscopic findings, with short fine telangiectasias. (D) After the excision of the largest tumor, hair in the area was shaved and up to 10 similar lesions of smaller size were observed.

After these histopathological findings, all the smaller papules that surrounded the biggest lesion were extirpated revealing the same histopathologic features. Because of the multiple lesions and the familiar history of malignancies, genetic counseling was given for the BAP1 tumor predisposition syndrome with a negative result.

Conclusions

BIMT is considered a type of intermediate melanocytic tumor, that has been recently described [1]. Clinically BIMTs are described as a unique skin-colored to reddish-brown, dome-shaped to pedunculated well-circumscribed papule, from 2 to 10 mm in diameter [2]. Dermoscopically BIMTs are characterized by homogenous pink or milky red background with sparse linear or arborising vessels [3]. Most commonly occur in the second and third decades of life [4].

The diagnosis is best confirmed by BAP1 immunochemistry with loss of nuclear expression in the altered melanocytes [1]. BAP1 is a tumor suppressor gene, and mutations of this gene result in some human cancers. Germline mutations have been described in families with a hereditary increased risk of certain cancers like cutaneous and uveal melanoma, malignant mesothelioma, renal cell carcinoma and other internal malignancies like lung or breast cancer [2,4].

Usually the proliferation index is low (<5%) so it is considered a low-grade dysplasia BIMT and the recommendation is to remove it completely with a 2 mm margin. If the proliferation is higher, it is considered a high-grade dysplasia BIMT and the suggested resection margins are from 5 to 10 mm [1]. Genetic counseling should be given according to personal and familial history of malignancies and in cases of multiples BIMTs [2,5].

BIMTs are considered a type of intermediate melanocytic tumor, typically described as a single erythematous papule. The presentation of multiple smaller-sized BIMTs surrounding a larger, more typical BIMT in the scalp is different to previously described cases.

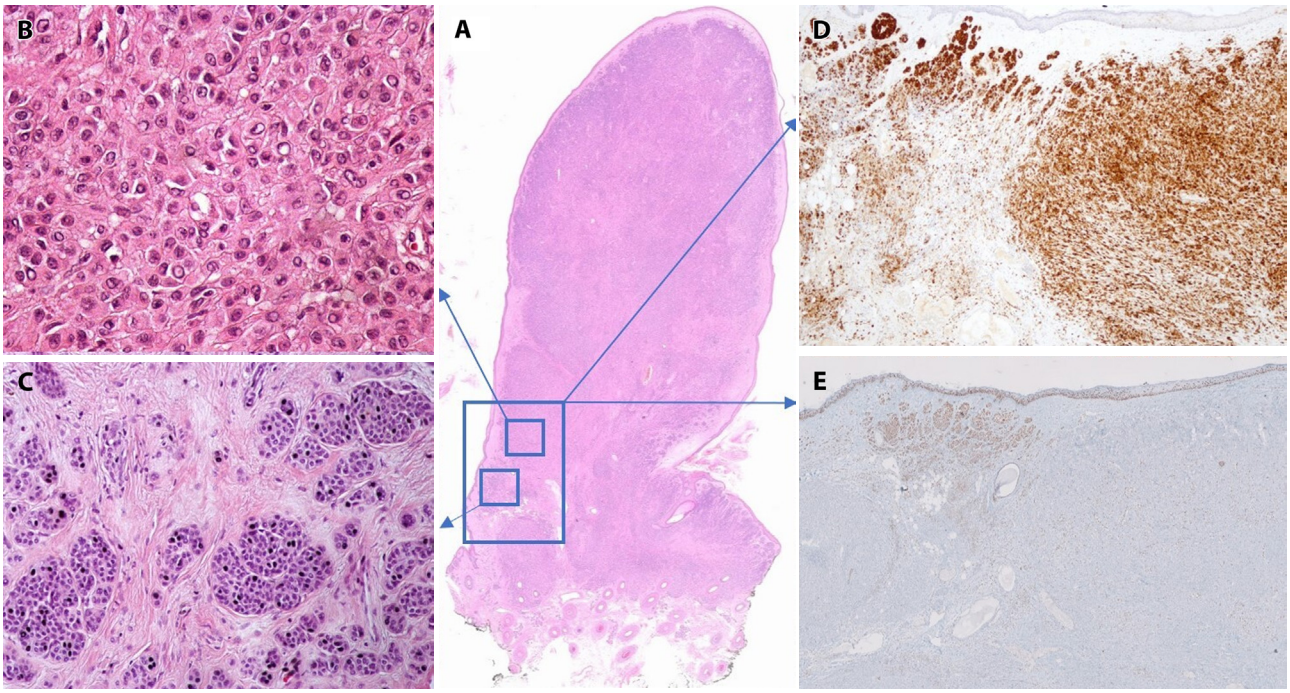

Figure 2. Histopathological findings. (A) The dermis was expanded by a cellular substitution arranged in nests and cords. (H&E, x 2). (B) Cells showed nevic appearance and epithelioid morphology (H&E, x 40). (C) It showed on the base a focus with nests of a conventional nevus (H&E, x 20). (D) Positive result for melan-A in the entire sample (IHC melan-A, x 10). (E) BAP1 expression was lost except in the conventional-appearing nevus component (IHC BAP1, x 2).

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