CASE REPORT

Atrichia with papular lesions in a Kuwaiti boy

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ABSTRACT

Atrichia with papular lesions (APL) is a rare autosomal recessive disorder resulting in complete and irreversible hair loss shortly after birth. Affected individuals also develop papular lesions of keratin-filled follicular cysts over extensive areas of the body. Mutations in the hairless gene have been shown to underlie the phenotype. We report a case of APL in a 13-year-old Kuwaiti boy. Increased recognition of this disorder will result in more accurate diagnosis and the sparing of unnecessarily treatment to patients.

KEY WORDS: Atrichia with papular lesions, hairless gene, alopecia totalis, alopecia universalis

INTRODUCTION

Atrichia with papular lesions (APL) (MIM 209500) is a rare form of total alopecia that is inherited in an autosomal recessive fashion.[1-3] The clinical symptoms in affected individuals include atrichia at birth or shedding of normal scalp hair several months after birth with failure to regrow; sparse eyebrow and eyelashes; and lack of secondary axillary, pubic or body hair. Histological examination of scalp biopsy is characteristically devoid of mature hair follicles.[4] Multiple follicular papules and morphologic and structural variations of follicular remnants that develop at approximately 2 years of age have been reported in APL patients.[1-4] These papules are distributed over different areas of the body, including scalp, cheeks, arms, elbows, thighs and knees. APL patients usually show normal growth and development, with normal teeth, nails, sweating, and hearing. APL was linked to chromosome 8, and subsequently, the human homologue of the murine hairless gene (HR) was identified as the gene responsible for APL. A series of pathogenic mutations have been identified in patients with APL around the world, thereby establishing the molecular basis of this disorder.[4-7]

CASE REPORT

A 13-year-old boy presented with alopecia universalis. It began shortly after birth with patchy alopecia that progressed to universal alopecia within 2-3 months. The patient developed scattered papular lesions when he was five years old. Family history and consanguinity were negative. Examination revealed universal hair loss (Fig. 1)

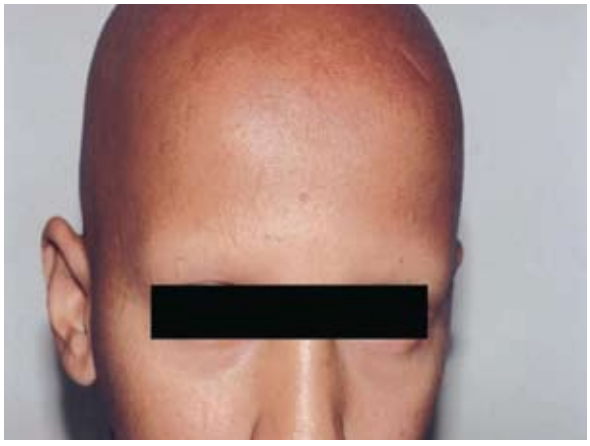

Fig. 1 Alopecia universalis.

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and scattered papular lesions some of which were milia-like while the others were rough-surfaced and skin-colored (Fig. 2). The teeth, nails and sweat glands were normal. The patient was physically as well as mentally normal.

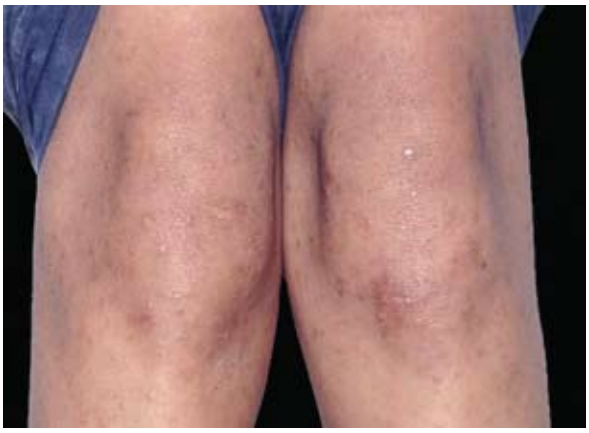

Fig. 2 Papular lesions over the knees.

His full blood count and blood biochemistry studies as well as thyroid function tests were normal. A biopsy specimen of the scalp showed absence of normal hair follicles with few hair follicles that were replaced by residual infundibulae (Fig. 3). Sebaceous and sweat glands were normal. Histological examination of a biopsy specimen taken from papular lesions of the knee revealed hyperkeratosis, acanthosis as well as follicular keratosis and keratinous cyst (Fig. 4). DNA studies proved extensive mutation of the hairless gene.

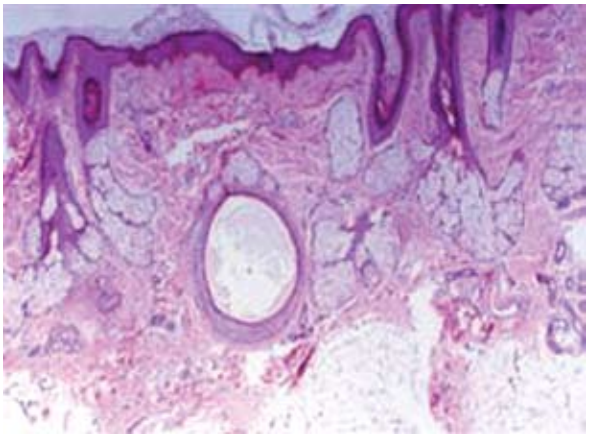

Fig. 3 Residual infundibulae and a follicular keratinous cyst are evident. Sebaceous and sweat glands are normal (hematoxylin and eosin, x40).

DISCUSSION

Atrichia with papular lesions (APL) (OMIM # 209500) is a rare form of irreversible alopecia that is inherited in an autosomal recessive pattern (Fredrich, 1950;

Damste and Prakken, 1954; Loewenthal and Prakken, 1961). In individuals affected with this form of hair loss, hairs are typically absent from the scalp, axilla and body. Patients are almost completely devoid of eyebrows and eyelashes. Histologic examination of affected scalp skin shows the absence of mature hair follicle structures. Although alopecia may accompany several different forms of congenital ectodermal dysplasias, APL patients are unique in that, along with total atrichia, papules and follicular cysts filled with cornified material represent a unique cutaneous abnormality among inherited alopecias. Zlotogorski et al have poroposed an algorithm of diagnostic criteria for congenital APL. Family history, especially of consanguinity, clinical history, scalp biopsy, and a mutation in the HR gene are all important considerations in diagnosing APL.[8] Interestingly in our patient family history and consanguinity were negative. However the criteria proposed by Zlotogorski et al are very useful in establishing

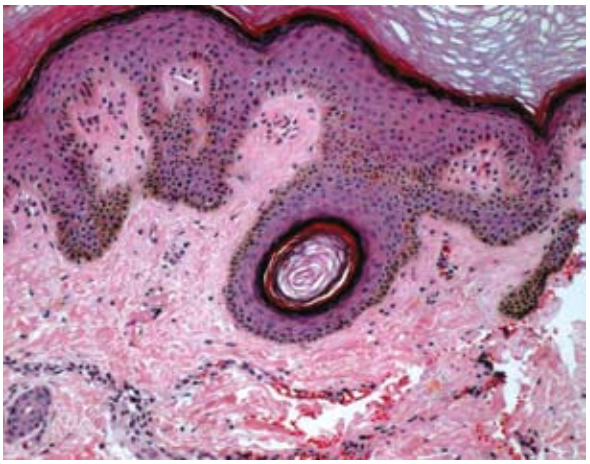

Fig. 4 Hyperkeratosis, acanthosis and keratinous cyst (hematoxylin and eosin, x100).

the diagnosis of APL since it could easily be dismissed by the clinician as a case of alopecia totalis alopecia universalis (AT/AU) and the associated papular rash could be attributed to a form of keratosis pilaris as an explanation for the papular lesions.[8] Probably the phenotype of atrichia with papular lesion is more common than was earlier believed, and in the near future, clinicians will discover some APL patients that were labeled and treated accordingly as AU. Therefore, increased recognition of this disorder will result in more accurate diagnosis and the sparing of unnecessarily treatment to patients.

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