Congenital atrichia with papular lesions

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Abstract
Congenital atrichia with papular lesions is a rare, autosomal recessive and irreversible form of total alopecia of the body hair characterized by hair loss soon after birth and the development of keratin-filled cysts or horny papules over extensive areas of the body. The condition is associated with a mutation of the human hairless gene on chromosome region 8p12. We report a 1-year-old boy presenting with the absence of scalp and body hair since birth. On examination, he had complete absence of hair on the scalp, eyebrows, and eyelashes. Multiple, discrete, pearly-to-skin-colored papules of 1-3mm in size were present over the scalp. The skin biopsy from a scalp papule revealed normal overlying epidermis with multiple keratin cysts and hypoplastic hair follicles in the upper dermis.

Keywords: congenital atrichia, papular lesions over scalp, cluster of white dots, keratin filled cysts, congenital alopecia

Introduction
Congenital alopecia includes congenital atrichia with papular lesions (APL), alopecia universalis, vitamin D-dependent rickets type IIA, and syndromes such as Moynahan syndrome, hidrotic ectodermal dysplasia, and progeria. Congenital atrichia with papular lesions is a rare, autosomal recessive and irreversible form of total alopecia of the body hair characterized by hair loss soon after birth associated with the development of keratin-filled cysts or horny papules over extensive areas of the body. A mutation of the human hairless gene on chromosome region 8p12 is responsible [1].

Case Synopsis
A one-year-old boy, born of a third-degree consanguineous marriage, was brought to the outpatient department of dermatology because of the absence of scalp and body hair since birth. At the age of 10 months, the parents also noticed multiple skin colored raised papules over the scalp, which progressively increased in number. He was previously treated for alopecia universalis and vitamin D-dependent rickets type IIA but his alopecia was unresponsive to therapy. Developmental milestones were within normal limits. Sweating and body temperature was normal. There was no history of decreased hearing or seizures. A 15-year-old relative of the patient reportedly has similar lesions (Figure 1).

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On examination, the patient had a complete absence of hair on the scalp, eyebrows, and eyelashes (Figure 2, 3). Multiple, discrete, pearly to skin colored papules of 1-3mm in size were present over the scalp. He had no abnormalities of nails, teeth, mucosae, palms, and soles. His physical growth was normal according to age. No bony abnormalities, dysmorphic features, or systemic involvement was present. Blood counts and liver and renal profiles were normal. The Vitamin D3 levels (1,25-dihydroxycholecalciferol) were 20.44ng/ml (normal: 30-100ng/ml). Radiographs of the wrist joints were normal. The diagnoses of congenital atrichia with papular lesions, alopecia universalis, and vitamin D-dependent rickets IIA were considered.

Trichoscopy (Mini 3000 LED Dermatoscope, Heine, Germany) showed pinpoint white dots arranged in clusters with no perifollicular pigmentation, inflammation, or occlusion (Figure 4). The skin biopsy from a scalp papule revealed normal overlying epidermis with multiple keratin cysts and hypoplastic hair follicles in the upper dermis (Figure 5). There were no terminal hairs or sebaceous glands. On the basis of clinical, trichoscopic, and histopathological findings we confirmed the diagnosis of congenital atrichia with papular lesions (APL).

Case Discussion

Congenital atrichia with papular lesions is a rare and irreversible form of total alopecia of the scalp, eyebrows, eyelashes, and axillary and pubic areas characterized by hair loss soon after birth and the development of keratin-filled cysts or horny papules over extensive areas of the body involving face, neck, limbs, and trunk [1]. It is inherited as an autosomal recessive disorder. This condition has been noted for decades among a group of people known as Irish Travelers, who have existed as a distinct indigenous
Congenital atrichia with papular lesions without ectodermal dysplasia is a very rare disorder and can be easily misdiagnosed as alopecia universalis or vitamin D-dependent rickets type IIA. However, trichoscopy and histopathological examination helped us to reach the final diagnosis of congenital atrichia with papular lesions. Hence, a high index of suspicion should be kept in mind in patients presenting with generalized alopecia to avoid misdiagnosis.

### Conclusion

Congenital atrichia with papular lesions without ectodermal dysplasia is a very rare disorder and can be easily misdiagnosed as alopecia universalis or vitamin D-dependent rickets type IIA. However, trichoscopy and histopathological examination helped us to reach the final diagnosis of congenital atrichia with papular lesions. Hence, a high index of suspicion should be kept in mind in patients presenting with generalized alopecia to avoid misdiagnosis.

### Potential conflicts of interest

The authors declare no conflicts of interests.
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