Case Report
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Atrichia congenita with papular lesions: A rare cause of pediatric alopecia

Papüler lezyonlu konjenital atrişi: Pediatrik alopesinin nadir bir nedeni

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Abstract

Congenital atrichia with papules is a rare inherited disorder characterized by the replacement of hair follicles with keratinous cysts, resulting in hair loss shortly after birth and papular lesions. It should be differentiated from other causes of congenital atrichia, such as vitamin D-dependent rickets, alopecia universalis, and ectodermal dysplasia, to avoid unnecessary administration of medications and to counsel parents regarding the benign but irreversible nature of the condition.
Keywords: Congenital alopecia, atrichia, papular

Öz

Papüler lezyonlu konjenital atrişi, saç foliküllerinin yerini keratinöz kistlerin alması, doğumdan kısa süre sonra gelişen yaygın saç dökülmesi ve papüler deri lezyonları ile karakterize, nadir görülen kalıtsal bir hastalıktır. Gereksiz tedavilerden kaçınmak ve ebeveynleri hastalığın benign ancak irreversibl olduğu konusunda bilgilendirmek amacıyla; bu durumun D vitaminine bağımlı raşitizm, alopesi üniversalis ve ektodermal displazi gibi diğer konjenital atrişi nedenlerinden ayırt edilmesi gerekir.
Anahtar Kelimeler: Konjenital alopesi, atrişi, papüler

Introduction

Atrichia congenita with papular lesions (APL) is a rare inherited disorder caused by mutations in the human hairless gene (HR) on chromosome 8p21-22[1]. It is clinically characterized by the irreversible loss of hair shortly after birth, associated with a diffuse papular eruption. Histologically, these papules represent follicular keratin-filled cysts formed due to an abnormal hair cycle[2]. Herein, we report a case of APL in an Indian girl.

Case Report

A six-year-old girl presented with complete absence of scalp and body hair along with multiple raised lesions involving the scalp, face, body, and limbs. The child exhibited normal hair growth at birth. Spontaneous hair loss began at the age of 2 months on the scalp and gradually progressed to her eyebrows and eyelashes. Multiple treatment attempts failed to induce hair regrowth. At the age of 4 years, the parents noticed the appearance of asymptomatic raised lesions on her scalp and face, which eventually spread to the rest of her body. She

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had no history of atopy, reduced sweating, hearing difficulty, blurred vision, seizures, or bone pain. She was the second child of a second-degree consanguineous marriage, born via Caesarean delivery after an uneventful pregnancy. She had attained developmental milestones at the usual age. Neither her elder sister nor her parents had any similar complaints.

Examination revealed a complete absence of hair throughout the scalp and body (Figure 1 and 2). There were multiple skin-colored to hyperpigmented discrete papules, ranging from 1 to 5 mm in size, distributed symmetrically over the scalp, face, trunk, and extremities, sparing the palms and soles (Figure 2). The nails, teeth, and mucosa did not show any abnormalities. Her physical development was normal for her age at presentation. There were no bony deformities or dysmorphic features, except for mild lateral protrusion of the ears. Differential diagnoses of APL, vitamin D-dependent rickets (VDDR) type IIA, alopecia universalis, and hidrotic ectodermal dysplasia were considered.

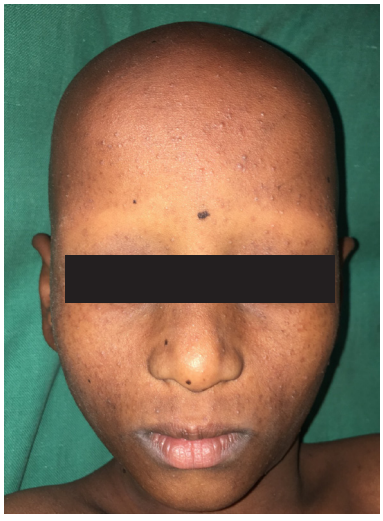

Figure 1. Complete loss of hair on scalp

Hematological, hepatic, and renal parameters, as well as calcium and phosphate levels, were within the normal range. The vitamin D3 level was 36.89 ng/mL (normal: 30-100 ng/mL). Radiographs of the bilateral wrist joints and lower limbs did not show any features suggestive of vitamin D deficiency. Thus, the clinical picture pointed towards a diagnosis of APL. However, histopathological evidence could not be obtained because the patient was unwilling to undergo a skin biopsy.

Discussion

APL is an autosomal recessive genodermatosis reported to be more prevalent in a minority gypsy population within Ireland, known as the Irish travelers[3]. The exact molecular pathogenesis of this condition is unknown. It is hypothesized that the HR gene encodes a transcription co-repressor factor protein that plays an important role in regulating catagen remodeling in the hair cycle. Mutations in this gene cause hair matrix cells to undergo premature and massive apoptosis and separate from the overlying epithelial sheath. Consequently, hair bulbs and dermal papillae become trapped in the dermis, leading to the cessation of hair growth and the formation of keratinous follicular cysts[4,5].

Although originally termed "congenital atrichia" by Ahmad et al.[3], these patients are usually born with normal hair, which is lost permanently within a year of birth. Another distinct feature of APL, as the name suggests, is the development of papular lesions that represent keratin-filled cysts. The lesions are usually generalized, as in our patient, although isolated involvement of the scalp has also been reported by previous authors[5,6]. On trichoscopy, lesions show pinpoint white dots representing cicatricial alopecia, but there are no signs of inflammation, perifollicular pigmentation, or follicular occlusion[6].

APL should be differentiated from alopecia universalis, which also presents with generalized non-scarring hair loss but lacks characteristic papular lesions. Ectodermal dysplasia may be excluded if other ectodermal components, such as teeth, nails, and sweat glands, are normal. VDDR type IIA often presents with congenital atrichia but can be ruled out in the presence of normal serum vitamin D3 and calcium

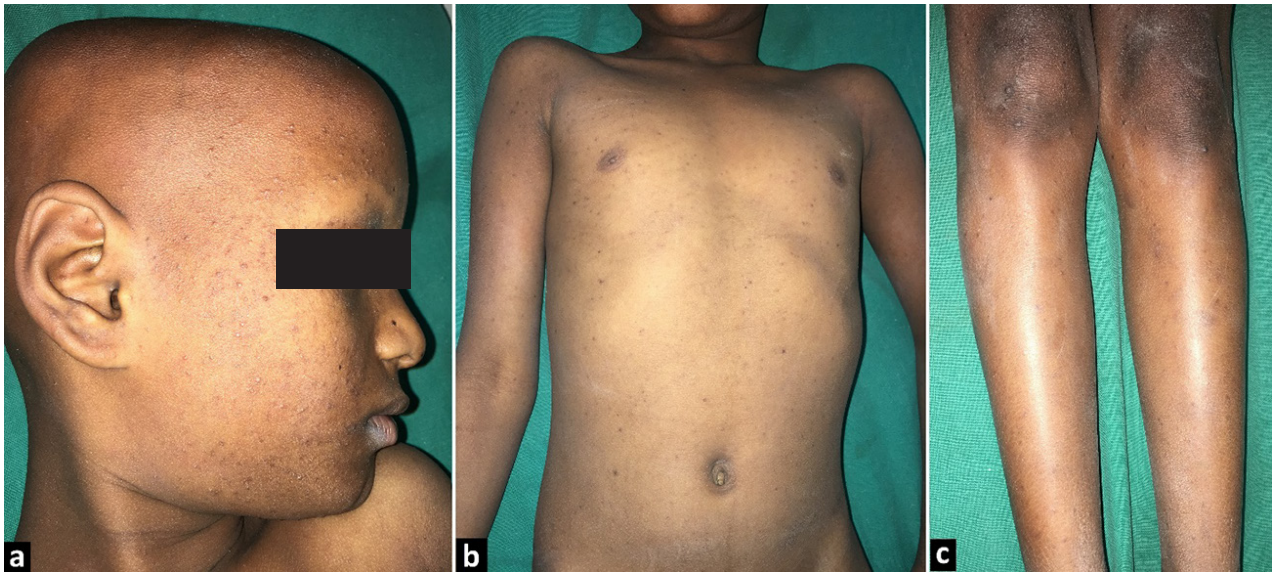

Figure 2. Discrete skin-colored to hyperpigmented papules distributed over the (**a**) scalp and face, (**b**) trunk and arms, and (**c**) legs

Table 1. Revised diagnostic criteria for APL[8]

Major criteria: (4 out of 5 required for diagnosis)
• Permanent and complete disappearance of scalp hair by few months of age. • Few or widespread smooth, whitish or skin-colored milia-like papules on the scalp, face, trunk and extremities from infancy or childhood. • Scalp histology showing replacement of mature hair follicles by follicular cysts filled with cornified material. • Mutation(s) in the human hairless gene detected through genetic testing. • Clinical and/or biochemical exclusion of vitamin-D-dependent rickets
Minor criteria: (supplementary criteria)
• Family history of consanguinity. • Absence of secondary axillary, pubic, or body hair and/or sparse eyebrows and eyelashes. • Normal growth and development, and normal bones, teeth, nails, and sweating. • Whitish hypopigmented streaks on the scalp. • Lack of response to any treatment modality.

levels and normal wrist joint radiographs, as seen in our case. Zlotogorski et al.[7] proposed two main laboratory findings for confirmation of APL diagnosis: i) histopathological evidence of replacement of the lower portions of hair follicles by keratin-filled cysts and ii) molecular evidence of mutations in the HR gene. Yip et al.[8] later published a set of revised diagnostic criteria, as listed in Table 1. Our patient met three major and four minor criteria. Various other conditions have been linked with APL, such as Moynahan syndrome (epilepsy, mental retardation), situs inversus, mesocardia, hidrotic ectodermal dysplasia and premature ageing syndromes[4,6,9]. However, no such association was observed in the present study.

The management of APL is difficult because of the lack of effective treatment options for this condition. Wigs and hair prostheses may be useful camouflage measures. In addition, the psychosocial impact of congenital alopecia is profound and should be adequately addressed in these patients, especially during their formative years[9].

In conclusion, APL is a rare genetic disorder that should be considered when evaluating a child with atrichia. It is important to rule out other conditions with a similar presentation to avoid unnecessary administration of medications and counsel the parents regarding its prognosis.

Ethics

Informed Consent: Written informed consent was obtained from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: B.K.K., A.D., N.R.N.G., S.C., Concept: B.K.K., N.R.N.G., S.C., Design: B.K.K., A.D., N.R.N.G., S.C., Data Collection or Processing: B.K.K., A.D., S.C., Analysis or Interpretation: B.K.K., A.D., N.R.N.G., Literature Search: B.K.K., A.D., N.R.N.G., S.C., Writing: B.K.K., A.D., N.R.N.G., S.C.

Conflict of Interest: No conflict of interest was declared by the authors.

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