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Case Report

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Papular atrichia

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ABSTRACT

Congenital atrichia or papular atrichia is a rare form of irreversible alopecia inherited autosomal recessively. Atrichia congenita with papular lesions represents a complex and heterogeneous group of genodermatoses characterized by irreversible complete hair loss soon after birth, and associated with the development of keratin-filled cysts over the body. Mutation of the human hairless (HR) gene on chromosome 8p22 has been implicated with this condition. It has broad differential diagnosis and poses diagnostic and therapeutic challenges. We report an 8 year girl presenting with complete loss of hair over scalp, eye brows, eye lashes, and body soon after birth with papular lesions over face.

Keywords: Atrichia congenita, Congenital atrichia with papules, HR

INTRODUCTION

Congenital alopecia also known as papular atrichia or Atrichia congenital with papular lesions is irreversible type of alopecia which is rarely encountered.[1] Atrichia congenita with papular lesions (APL) represents a heterogeneous group of genodermatoses characterized by irreversible complete hair loss soon after birth which is associated with the development of keratin-filled cysts over the body.[2,3] The condition may occur in isolation in both sporadic and familial forms or along with other defects In the isolated familial form, inheritance is usually autosomal recessive, although autosomal dominant inheritance has occurred in some families.[1,2] The gene locus for familial cases is on chromosome 8p21-22 (ALUNC-alopecia universalis congenitalis) and mutation of the human hairless (HR) gene has been proposed in atrichia congenital.[2,3] Here we report a child with congenital atrichia and papular lesions, which is very rarely reported in the literature.

CASE REPORT

An 8 year girl presented with complete loss of hair on the body soon after the birth. She had sparse hairs over the scalp at birth but gradually lost all of them within few months of birth. There was history of consanguinity (grade 2) in her parents. She had consulted various doctors and had received topical and oral medications but was unresponsive to therapies. She developed multiple white tiny lesions on the face and neck, which progressively increased in number since 2 years. Examination revealed multiple milia and skin colored papular lesions over the face and neck (Figure 1) and near complete absence of scalp, facial, axillary, and pubic hair (Figure 2). Physical growth was normal. Her teeth, nail, mucosa, palms, and soles were normal. There was no any systemic involvement and bony abnormality. Serum vitamin D3 level and calcium were normal. Radiographs of the wrist joint were normal.

Figure 1: Shows milia like papules and multiple skin coloured papules over the face (blue arrow) with near complete loss of scalp and eyebrow hair (star).

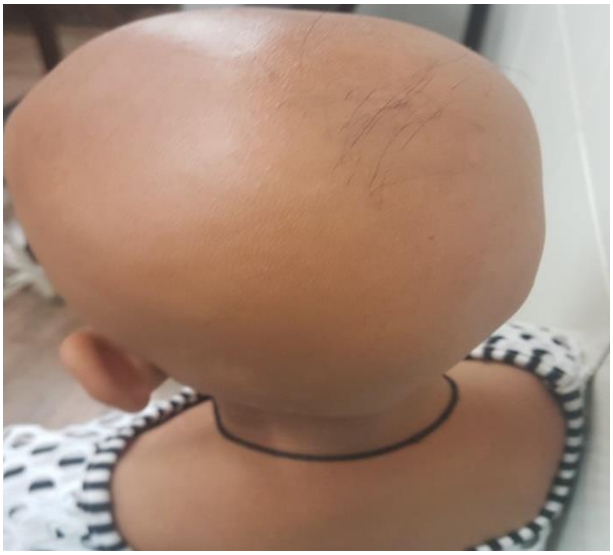

Figure 2: Near complete loss of hair over scalp.

DISCUSSION

Atrichia congenita with papular lesions is a rare, autosomal recessive form of total alopecia of the scalp, eyebrows, eyelashes, axillary and pubic hair, 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.[1,2] This condition was noted among the Irish Traveler gypsies. Ahmad et al first referred to this condition as congenital atrichia.[1] The condition is confused with congenital alopecia universalis, vitamin D dependent rickets, and ectodermal dysplasia. These patients have normal development, hearing, teeth and nails.[2] There are no abnormalities of sweating. Heterozygous individuals have normal hair and are clinically indistinguishable from genotypically normal persons.[2]

The exact molecular basis of this disease is not known. Mutations in the human HR located on chromosome 8p21.2 have been implicated.[1,3] It encodes for a putative single zinc-finger transcription factor protein, believed to regulate catagen remodeling in the hair cycle. It is hypothesized that the vitamin D receptor and HR genes, which are both zinc-finger proteins, may be in same genetic pathway controlling postnatal hair cycle.[1]

The histology from a papule shows multiple mid-dermal keratin cysts with granular layer and laminated keratin in the mid-dermis with sparse peri-follicular lymphocytic infiltrate.[3]

Zlotogorski et al proposed diagnostic criteria for atrichia congenita with papular lesions but later Yip et al revised them.[4] Our patients met two out of five major criteria supplemented by four minor criteria for the diagnosis of isolated form atrichia congenita.

Congenital atrichia associated with situs inversus and mesocardia has been reported. Other associations are Moynahan's syndrome, hidrotic ectodermal dysplasia and aging syndromes. Congenital atrichia without ectodermal dysplasia is very rare. Our case of congenital atrichia with papular lesions is very rarely reported entity in literature and is reported because of its rarity.[1]

Diagnostic criteria for atrichia congenita with papular lesions (revised by Yip et al)[4]

Major criteria

Permanent and complete absence of scalp hair by the first few months of life. Few to widespread smooth, whitish, or milia-like papules on the face, scalp, arms, elbows, thighs or knees from infancy or childhood. Replacement of mature hair follicle structures by follicular cyst filled with cornified material in scalp histology. Mutation (s) in the human hairless gene through genetic testing. Clinical and/or molecular exclusion of vitamin D dependent rickets.

Minor (supplementary) criteria

Family history of consanguinity. Absence of secondary axillary, pubic, or body hair growth and/or sparse eyebrows and eyelashes. Normal growth and development, including normal bones, teeth, nails, and sweating. Whitish hypopigmented streaks on the scalp. Lack of response to any treatment modality.

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