Atrichia congenita

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ABSTRACT

Atrichia congenita is a rare genodermatoses is characterized by a mutation of the human hairless (HR) gene on chromosome 8p22. There is loss of scalp hair between one to six months of age, after which no growth occurs. Eyebrow, eyelash, and body hair may also be sparse or absent; patients may have a few pubic and axillary hairs. The condition may present in isolation or along with other defects.

Key words: Atrichia Congenita, autosomal recessive, human hairless gene, loss of scalp hair

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INTRODUCTION

Atrichia congenita without ectodermal defects (isolated form) is a rare autosomal recessive condition characterized by the shedding of scalp hairs between one and six months of age, after which no growth occurs. Eyebrow, eyelash and body hair may also be sparse or absent; patients may have a few pubic and axillary hairs. It may occur either in isolation or with associated defects. Isolated congenital alopecia has been reported to occur in both sporadic and familial forms. In the isolated familial form, inheritance is usually autosomal recessive, although dominant or irregular 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 on chromosome 8p21-22 produces the clinical picture of atrichia congenita.[3,4] Human HR gene is a homologue of the murine hairless gene[3-5] and encodes a zinc-finger transcription factor protein that is expressed in the brain and the skin.

CASE REPORT

A one-and-a-half-month-old male baby was brought by his maternal grandmother with complaints of frontoparietal scalp hair recession, sparse eyelashes and eyebrows [Figure 1]. The grandmother as well as two of her siblings (one female and one male) all had absent scalp, facial, axillary, and pubic hair since infancy. All three had normal hair growth at birth that shed within a few months. Two other siblings had normal hair. Their condition was unresponsive to steroid therapy. Physical growth was normal. Teeth, nail, mucosa, palms, and soles were normal. There was no bony abnormality or any systemic involvement. Examination revealed complete absence of scalp, facial, axillary, and pubic hair with no papular skin lesions. There was no visible perifollicular inflammation. Serum Vitamin D3 level and calcium were normal. Radiographs of the wrist joint were normal.

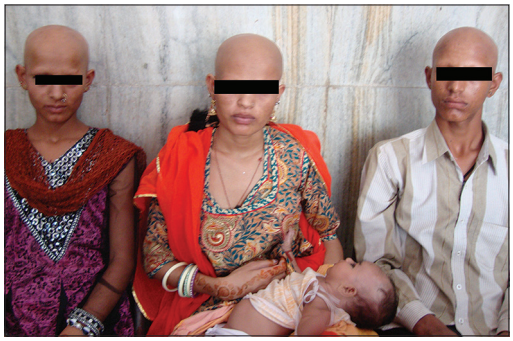

Figure 1: Three siblings (two females and one male) with absence of scalp, facial, pubic and axillary hair, along with the maternal grandson (one-and-a-half-months) having frontoparietal recession, sparse eyelashes and eyebrows

There was no history of delayed milestones in the baby, hypohidrosis, bone pain, hearing loss, or seizures. Examination revealed frontoparietal scalp hair recession, with sparse eyelashes and eyebrows. The baby was otherwise normal.

Table 1: Diagnostic criteria for atrichia congenita with papular lesions

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 dependant 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

DISCUSSION

Atrichia congenita (isolated form) is a rare autosomal recessive form of alopecia. Patients are born with hair that falls and is never replaced. The condition may be confused with congenital alopecia universalis, vitamin D dependant rickets, and ectodermal dysplasia. In our patients, congenital alopecia universalis was ruled out on the basis of lack of any history of sudden, patchy loss of normally appearing hair progressing to the loss of scalp, body, eyelash, andv eyebrow hair. Vitamin D dependent rickets was ruled out on the basis of lack of any history of joint pain, normal serum vitamin D3 and calcium levels, and a normal wrist joint radiograph. Ectodermal dysplasia was excluded as there was no history of delayed milestones or loss of sweating. Teeth, nails, mucosa (ectodermal components), palms, and soles were normal. The presence of a family history, history of consanguinity, and sparse eyebrows and eyelashes at the time of birth pointed to the diagnosis of atrichia congenita.

Zlotogorski *et al*. proposed diagnostic criteria for atrichia congenita with papular lesions but later Yip *et al*. revised them [Table 1].[6] Our patients met two out of five major criteria supplemented by four minor criteria for the diagnosis of atrichia congenita (isolated form).

The case is being reported for its rarity.

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