



CONGENITAL ATRICHIA- A RARE CASE REPORT

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ABSTRACT

Congenital atrichia is an autosomal recessive disorder. It is a rare form of irreversible alopecia. Atrichia congenita with papular lesions (APL) represents a complex and heterogeneous group of genodermatoses which is characterized by irreversible complete hair loss which occurs soon after birth, and associated with the development of keratin-filled cysts over the body. Homozygous mutation in the hairless gene (HR) is implicated. We report a case of congenital atrichia with papular lesions.

Key words: Congenital atrichia, Total alopecia, Atrichia congenita with papular lesions.

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INTRODUCTION

Atrichia congenita without ectodermal defects, is a rare autosomal recessive form characterized by shedding of scalp hairs between one to six months of age, after which no growth occurs. It may occur either in isolation or with associated defects. Isolated congenital alopecia has been reported to occur in both sporadic and familial forms. The gene locus for familial cases is on chromosome 8p21-22 and mutation of the human hairless gene (HR) on chromosome 8p21-22 produces the clinical picture of atrichia congenita.

CASE REPORT

A 4-year-old girl born to non-consanguineous parents was brought to the skin OPD with complaints of complete absence of hair on scalp and body from birth. (Fig 1&2). History from the mother revealed that the child was delivered by full

term, normal vaginal delivery with birth weight of 2.5kg. Antenatal and perinatal period was uneventful. Social and motor milestones were normal. No history of seizures, hearing or visual disturbances was present. The child has been vaccinated up to age. Child's sibling was asymptomatic.

At 1 1/2 years of age, mother noticed mild scanty hair over the scalp, which according to her parents was lost in 20 days. At 3 years of age, she started noticing multiple skin colored raised lesions over the scalp, back, hands and thighs. There was no history of decreased sweating, atopy or bone pain. Bowel and bladder habits were normal. There was no history of similar complaints in the family. Dermatological examination revealed complete loss of hair on the body, scalp and eyebrows. Multiple hyperkeratotic papules of size 3-5mm were present over the scalp, nape of the neck, back, B/L forearms and B/L thighs. Increased follicular prominence was observed over the scalp. Systemic examination was normal. The palms, soles, hair, nails, teeth and mucosa were normal. No bony abnormalities or dysmorphic features.

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HISTOPATHOLOGICAL EXAMINATION

Skin biopsy of lesion from the scalp revealed – Hyperkeratotic epidermis with minimal rudimentary

hair follicles, and lymphocytic infiltration seen in the superficial dermis (FIG 3). Hence we concluded to the diagnosis of congenital atrichia.

Figure 1. Clinical photograph showing multiple grouped hyperkeratotic papules present over the scalp.

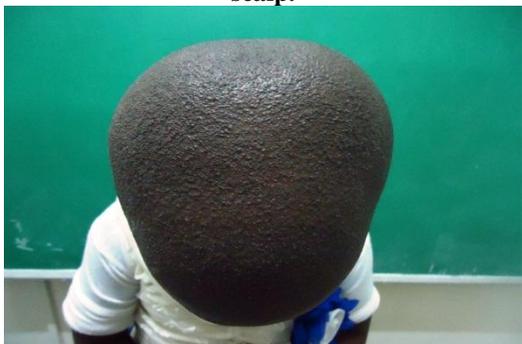


Figure 2. Clinical photograph showing multiple grouped hyperkeratotic papules present over the hand.



Figure 3. Histopathology slide from the scalp, revealed hyperkeratotic epidermis overlying pilosebaceous elements and hair follicles in the dermis. Hair follicles show presence of hair shafts. Mild lymphocytic infiltrate is seen in the superficial dermis.

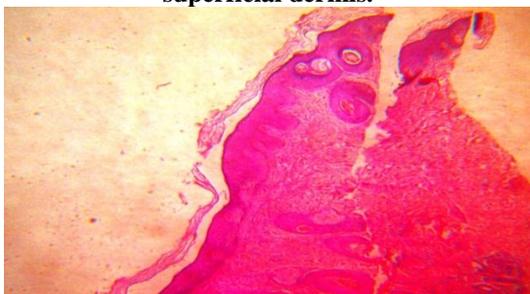
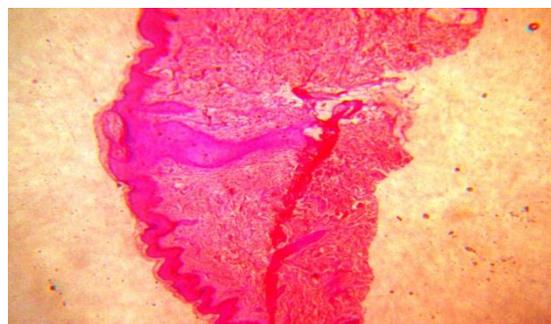


Figure 4. Histopathology slide from the left, arm shows epithelium overlying fibrocollagenous dermis. Superficial dermis shows lymphocytic infiltration, deep dermis showing presence of hair follicles.



DISCUSSION

Congenital atrichia is the complete loss of hair from birth. It can be inherited as autosomal recessive or autosomal dominant or X linked pattern. The cases inherited as autosomal recessive are the most severe form and are present since birth. Congenital atrichia usually presents as total alopecia at birth, but in some cases it can also present with scalp hair which is later shed between the first few months, after which no further regrowth occurs. It is associated with alopecia of eyebrows, eyelashes and general body hair. Congenital atrichia may be associated with papular leisons, which is a rare form of autosomal recessive syndrome, characterized by numerous, small, horny papules appear, first on the face, neck and scalp and then gradually progressing to involve the limbs. Histologically the papules are thick walled keratin cysts.

Nails, teeth, sweating, growth and development were normal. Histology showed total absence of hair follicles or a few miniaturized

follicles. Underlying disorder in papular atrichia, appears to be that towards the end of the first anagen phase the hair bulb, proximal inner root sheath and outer root sheath undergo premature and massive apoptosis and disintegrate into separate cell clusters that lose contact with the dermal papilla. As a result, the dermal papilla fibroblasts fail to migrate upward and break up into clusters shrunken cells stranded in the reticular dermis as dermal cyst precursors, and the follicle loses the ability to cycle. The exact molecular basis of this disease is unknown. Mutations in the human HR gene located on chromosome 8p21.2 have been implicated. It encodes for a putative single zinc-finger protein, believed to regulate catagen remodeling in the hair cycle. It is hypothesized that vitamin d receptor and HR genes, which are both zinc finger proteins, may be in same genetic pathway controlling postnatal hair cycle. Viamin D dependent rickets type II A induced alopecia and alopecia univalaris are important differential diagnosis for APL. Syndromes associated with congenital atrichia

are hidrotic ectodermal dysplasia (total or almost total alopecia, palmoplantar keratoderma, and thickened nails), Moynahans syndrome (mental retardation, epilepsy) and aging syndromes.

Diagnostic criteria for APL involve:

1. Family history
 - a. pattern of inheritance
 - b. history of consanguinity
2. Examination
 - a. complete lack or almost complete lack of scalp hair
 - b. sparse eyebrows & eyelashes
 - c. lack of secondary axillary, pubic hair.
 - d. papules distributed over the face, scalp and extremities
3. Biopsy
Absence of matured hair follicles and cysts filled with cornified material.
4. Mutation in the hr gene.
Our patient is a case of congenital atrichia with papular leisons.

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CONCLUSION: We report a case of congenital atrichia with papular leisons because of its rare occurrence and have to be differentiated from alopecia universalis. Hence this disorder may be diagnosed accurately and spared the battery of treatments that are destined to fail in APL.

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CONFLICT OF INTEREST:

The authors declare that they have no conflict of interest.

STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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